VDH VIRGINIA DEPARTMENT OF HEALTH

Division of Child and Family Health- Newborn Screening Follow-Up Program

Spinal Muscular Atrophy (SMA)

Parent Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose your baby with the condition. Many babies who receive abnormal results do not have the condition. Follow-up with your provider is **very important** to determine if your baby has the disorder indicated.

Disorder Indicated: Spinal muscular atrophy genetic disease that affects the motor neurons in the spinal cord. Motor neurons are specialized nerve cells that control muscles throughout the body including those used for chewing/swallowing, breathing, and movements of the arms and legs. Spinal muscular atrophy leads to loss of motor neurons and results in progressive weakness of muscles without treatment.

Children with SMA who are identified through newborn screening will have better outcomes from early detection and diagnosis. Clinical trial data has demonstrated improved motor function and survival with early institution of treatment. Urgent evaluation and follow-up is critical.

Incidence	1 in every 10,000 newborns.
Signs and Symptoms *Please note: these findings may not be present in young infants or in milder forms of the disease*	Individuals with spinal muscular atrophy have a missing or mutated SMN1 gene. This is the gene responsible for creating a protein that maintains the health of the motor nerves. Without normal SMN protein, the motor nerves degenerate, leading to weakness in muscles throughout the body. The severity of weakness in a patient with SMA varies. Patients may be grouped based on their maximal motor milestones accomplished, such as the ability to sit, stand, or walk. These abilities will be impacted by treatment. *A child with spinal muscular atrophy may demonstrate symptoms including:* • Poor head control • Weakness, more prominent in the legs compared to arms (with reduced kicking or difficulties reaching for toys) • Absent reflexes • Delays in motor milestones, or loss of previously gained motor skills • Low muscle tone (hypotonia) • Weak cry • Poor feeding leading to failure to thrive (lack of appropriate weight gain) • Respiratory insufficiency leading to respiratory distress with illness or abnormalities of chest wall appearance including a bell-shaped chest *(Of note, SMA is not known to impact cognitive or social development, with most babies demonstrating alert, attentive, interactive abilities)*
Next Steps ***may*** include:	**Follow up with your child's pediatric provider** **Possible referral to pediatric neuromuscular specialist/SMA Care Center** Clinical assessment Genetic Testing
Treatment ***(if indicated)***	There are medications available for the treatment of SMA, as well as ongoing clinical trial opportunities to evaluate new treatments
Additional Resources	VDH Newborn Screening VDH Newborn Screening *www.vdh.virginia.gov/newborn-screening/* Baby's First Test *www.babysfirsttest.org* American College of Medical Genetics (ACMG) ACT Sheets *www.ACMG.net* Genetics Home Reference *https://ghr.nlm.nih.gov/* Cure SMA *http://www.curesma.org/* SMA Foundation *http://www.smafoundation.org/* Muscular Dystrophy Association *http://MDA.org/*

Educational content adapted from www.babysfirsttest.org

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