VDH VIRGINIA DEPARTMENT OF HEALTH

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

Spinal Muscular Atrophy (SMA)

Health Care Professional Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An "abnormal" or "critical" result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

Disorder Indicated: Spinal muscular atrophy (SMA) is a genetic disease that affect the motor neurons of 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 and muscle atrophy without treatment. Children with SMA who are identified through newborn screening may have better outcomes through early intervention and treatment. Timely follow-up is critical for diagnosis and treatment.

Incidence	1 in every 10,000 newborns.
Analyte Measured	Homozygous Exon 7 deletion in SMN1 gene (Real-Time PCR Assay)
Normal Test Results	SMN1 Exon 7 Amplification within NORMAL Range
Abnormal Test Results	SMN1 Exon 7 Amplification within ABNORMAL Range
Critical Test Results	SMN1 Exon 7 Amplification within CRITICAL Range **(Critical results require immediate evaluation and follow-up)**
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 unifying genetic etiology of a missing or mutated SMN1 gene. This is the gene responsible for creating a protein that maintains the health of the motor neuron. In the absence of appropriate SMN protein production, the motor neurons deteriorate leading to weakness in muscles throughout the body. The phenotype, or severity of weakness, of a patient with SMA varies. Individuals are characterized based on their maximal motor milestones accomplished, such as the ability to sit, stand, or walk. The most common phenotype is the most severe, with weakness usually in the first few months of life. Without treatment, these babies will not survive past age two years. *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:	**Discuss the next steps of evaluation and urgent referral to Regional Pediatric Neuromuscular/SMA Care Center** Provide parental education (see accompanying sheet) Clinical assessment Genetic Testing
Treatment ***(if indicated)***	FDA approved medications, Gene Therapy, and/or participation in clinical trial opportunities
Additional Resources	VDH Newborn Screening *www.vdh.virginia.gov/newborn-screening/* SMA Educational Module *https://smartmoves.curesma.org/* 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

VIRGINIA NEWBORN SCREENING PROGRAM

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