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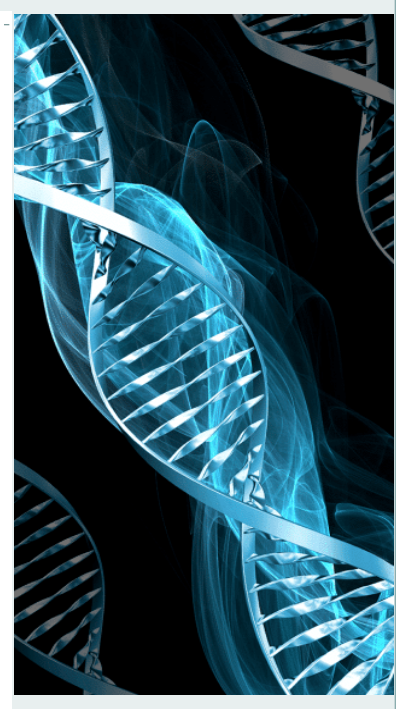

Spinal Muscular Atrophy to be Added to Nebraska's Newborn Screening Panel

Beginning November 14, 2020, Nebraska's newborn screening panel will grow to 33 rare diseases. Babies born on or after November 14, 2020, will be screened for Spinal Muscular Atrophy (SMA). Learn more about this condition in this issue.

ABOUT SMA

Geetanjala Rathore, MD Pediatric Neurologist, Children's Hospital Omaha

Spinal muscular atrophy (SMA) is a genetic neuromuscular disorder that presents with weakness of the skeletal and respiratory muscles. SMA affects approximately 1 in 10,000 births, and about 1 in every 50 Americans is a carrier for this disorder. SMA is caused by a mutation in the survival motor neuron gene 1 (SMN1). In unaffected individuals this gene produces a protein called survival motor neuron (SMN) protein, which is critical to the function of motor neurons in the spinal cord. In the absence of this protein, the motor neurons cannot properly function and die, leading to debilitating and often fatal muscle weakness.

A second gene, called survival motor neuron gene 2 (SMN2), is like a backup gene. It codes for a modified SMN protein, which lacks a key building block that is normally produced by SMN1. Patients with SMA can have anywhere from 1 to 5 or even more copies of SMN2. The more copies of SMN2 a patient has, less severe the form of SMA presents as.

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We extend our gratitude to Geetanjala Rathore MD, Pediatric Neurologist for her contributions to this update.

ABOUT SMA

According to the SMA Foundation, SMA is the # 1 genetic killer of infants and toddlers.

The more copies of SMN2 a patient has, less severe the form of SMA presents as. There are five primary types of SMA: Type 0, 1, 2, 3, and 4. The type of SMA is based on the onset and highest physical milestones achieved.

Type 0: Type 0 is both rare and severe. Symptoms begin prior to birth. Decreased fetal movement may be observed in the weeks prior to delivery. At birth, the infant has severe weakness and often difficulty breathing, feeding, and may have joint contractures and cardiac defects. These infants typically require respiratory and feeding support prior to confirming the diagnosis.

Type 1: Type 1, also known as Werdnig-Hoffman disease, is the most common. Without screening, diagnosis is often made during an infant's first 6 months of life. Babies with SMA Type 1 have significant muscle weakness, trouble breathing, coughing, and swallowing. They may need breathing assistance or a feeding tube. If not treated, Type 1 can be fatal within the first 2 years of life.

Type 2: Children with this type typically have delayed motor milestones and display a range of physical disabilities. Generally individuals affected by SMA Type 2 can sit up without support; however, some may need assistance getting into a seated position.

Type 3: Without screening Type 3, also called Kugelberg-Welander disease or juvenile SMA, is usually diagnosed after 18 months of age, and before 3 years of age. SMA Type 3 can be diagnosed as late as the teen years. Individuals affected by SMA Type 3 often have regression of motor skills they previously achieved.

Type 4: The onset of Type 4 is usually in adulthood, and it leads to mild motor impairment. While symptoms can begin in early adulthood, they often appear later in life. This is a less common form of SMA.

The diagnosis of SMA is based on molecular genetic testing. Genetic testing of SMN1/SMN2 is highly reliable. The absence of both full SMN1 copies will provide diagnosis of SMA.

These patients benefit the most from a multidisciplinary care approach. Their breathing, nutrition, range of motion, musculoskeletal integrity all need to be monitored and managed closely. They require regular physical and occupational therapy and orthotics. There are three FDA approved treatments available that are potentially lifesaving options for these patients. Nusinersen (Spinraza), requires intrathecal infusions every four months. Nusinersen increases the SMN1 production and improves the function of the existing SMN 2 gene production. The second therapy is gene replacement, Zolgensma, which is a one-time intravenous infusion which replaces the missing SMN1 gene and potentially cures the patient. The most recent therapy is an oral treatment, Evrysdi (Risdiplam) which helps to produce and maintain the existing SMN protein.

Based on research and clinical experience, early treatment of SMA leads to the best outcomes. Pre-symptomatic diagnosis and treatment has shown normal development in these patients who would not have survived beyond the age of 2 years. The screening of every newborn for this disorder can prevent deaths or the need for permanent ventilation in infants less than 1 year old with SMA. It is critical to diagnose these babies early and start treatment immediately.

Reminder:

Newborn Screening (bloodspot) is mandatory for all babies born within the state of Nebraska. The law is in place to protect infants from diseases that can't be easily identified without the test, and is required only for diseases that can be treated to help prevent infant mortality and reduce morbidity.

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NEBRASKA NEWBORN SCREENING PROGRAM

The mission of the Nebraska Newborn Screening Program is to prevent or minimize morbidity and mortality through newborn screening. The staff of the program are here to assist you with any questions:

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