EARLY ACTION, EARLY TREATMENT, SAVES LIVES.

Spinal muscular atrophy (SMA) is a rare autosomal recessive neuromuscular disease caused by deletion/mutation in SMN 1 gene, which is critical to the development of muscle mass and strength. It is characterized by progressive hypotonia and weakness. SMA puts a significant financial, logistical, and emotional strain on affected individuals and families.[1,2] Literature reviews indicate a significant diagnostic delay in SMA, during time periods that overlap with major motor neuron loss. For instance, in individuals affected by SMA Type 1, the average age of diagnosis is documented to occur between 5.3 to 6.3 months.[2,3] Yet, the onset of irreversible denervation occurs within the first 3 months, with loss of 90 percent of motor units occurring within 6 months of age.[4] However, clinical and preclinical studies indicate that treatment exposure early is critical to modifying the rapid and irreversible loss of motor neurons.[5-8]

The diagnosis of SMA, especially Type 1, is a medical emergency. To promote reduction of diagnostic delays, Cure SMA launched SMArt Moves, a disease awareness and educational campaign to empower parents, pediatricians, and other healthcare professionals to promptly recognize and diagnose the early signs of SMA.

Central to the SMArt Moves effort is a special section dedicated to healthcare professionals, detailing current diagnostic criteria, educational resources, and the latest treatment options and protocols. Available resources include:

- **SMA Diagnostic Toolkit:** Summarizes clinical trial data supporting early treatment, provides a table of clinical signs and symptoms by SMA type, and features a list of disorders to consider in the differential diagnosis of SMA
- **SMA 1-Page Quick Reference Guide:** An abbreviated version of the SMA Diagnostic Toolkit.
- **Know the Warning Signs:** A series of videos that break down some of the hallmark symptoms of SMA.
- **SMA CME HubSpot:** Free and accredited SMA related activities.

In addition, parents also have access to an easy-to-use website that encourages parents to trust their instincts if they suspect a motor delay, because missed milestones may be a sign of a serious medical condition like SMA. On the site, parents improve their understanding of the early signs of motor delays, watch instructional videos, and download a helpful checklist to share with their doctor and help address their concerns.

What About Newborn Screening?

Cure SMA recognizes that early diagnosis and treatment occurs most effectively with universal newborn screening. SMA was added to the Recommended Uniform Screening Panel (RUSP) in July 2018. As each state works to implement SMA screening within their newborn screening panel, providers must continue to be vigilant for the early signs to optimize outcomes.

Additionally, even in states in which SMA has been added to the panel, we encourage providers to remain watchful, as approximately 3% to 5% of individuals with SMA will not be identified by newborn screening due to SMN1 point mutations. Thus, clinical evaluation and consideration of the SMA diagnosis will remain important once universal inclusion of SMA within newborn screening panels is achieved.

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SMART MOVES

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