

Dear Primary Care Provider,

Spinal Muscular Atrophy (SMA) is a genetic neuromuscular disease characterized by weakness of the skeletal and respiratory muscles. SMA is a rare disorder occurring approximately 1 in 11,000 births, and about 6 million Americans are genetic carriers. SMA is caused by a missing gene called the Survival Motor Neuron 1 (SMN1); this is the gene that encodes the SMN protein. The SMN protein is critical to the health and survival of the nerve cells in the spinal cord responsible for muscle contraction (motor neurons). There is a wide range of severity, from type I in which patients cannot sit independently and require aggressive interventions for survival, to type II and III patients in whom the care needs are variable depending on the severity of their weakness.

Patients with SMA often have impaired cough, respiratory insufficiency, dysphagia, gastroparesis, constipation, and evolving orthopedic issues including scoliosis. Various types of equipment may be used, from respiratory support during sleep, e.g. BiPAP and mucus clearance devices, to gastrostomy tubes, to wheelchairs, and braces. Cognitive development is usually not affected. Despite these specialized needs to optimize function, you, the primary care provider, remain an integral part of the care team. Usual primary care practice, especially care coordination, family support, as well as routine pediatric care immunizations, developmental surveillance, and monitoring of growth contribute to the overall well-being of this child and their family.

Cure SMA offers many documents and resources addressing the standard of care for patients with SMA, including our SMA Care Series Booklets which can be used with clinicians and families. **Please email [infopack@curesma.org](mailto:infopack@curesma.org) or call 800-886-1762 with your mailing address to receive these publications.** Cure SMA maintains a website with many resources for both families and healthcare professionals visit us at [www.curesma.org](http://www.curesma.org). Should any clinical issues arise the members of the Medical Advisory Council are available to you. We represent a number of disciplines, including neurology, pulmonology, critical care, rehabilitation medicine, orthopedics, physical therapy, nutrition, psychology, primary, and palliative care.

Although challenging, we have found caring for patients and families with SMA to be a very rewarding experience. Please let us know what we can do to help.

Sincerely,

The Cure SMA Medical Advisory Council