

SMA: Survival isn't the only goal anymore

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Spinal muscular atrophy (SMA) was once a devastating diagnosis, often leading to severe disability and early mortality. The advent of disease-modifying therapies has transformed the natural history of SMA, allowing many patients, particularly those treated pre-symptomatically or in early infancy, to reach developmental milestones previously unimaginable. This success, however, has created a new clinical reality: a growing cohort of SMA patients is now reaching adulthood, presenting clinicians with novel challenges in managing their long-term health and quality of life.

KEY TAKEAWAYS

- **The Pivot:** Early intervention for SMA has shifted the clinical focus from survival to long-term adult care, including managing chronic complications and promoting independence.
- **The Data:** While specific long-term outcome data for adults treated from infancy is still emerging, existing therapies have demonstrated significant improvements in motor function and survival in pediatric populations.
- **The Action:** Clinicians must develop integrated, multidisciplinary care models tailored to the unique needs of adult SMA patients, addressing not only motor function but also respiratory, nutritional, skeletal, and psychosocial aspects.

Spinal muscular atrophy is a severe, progressive neuromuscular disorder caused by a deficiency of the survival motor neuron (SMN) protein, essential for motor neuron function.¹ The disease spectrum ranges from severe, infantile-onset SMA Type 1, where infants rarely survive beyond two years without intervention, to milder adult-onset forms.¹ The genetic basis involves homozygous deletion or mutation of the SMN1 gene, with disease severity modulated by the number of copies of the paralogous SMN2 gene. Lower SMN protein levels lead to motor neuron degeneration, progressive muscle weakness, and atrophy.

Before the introduction of disease-modifying therapies, standard care for SMA was largely supportive, focusing on managing symptoms such as respiratory insufficiency, feeding difficulties, and musculoskeletal complications.¹ This included respiratory support, nutritional interventions, and orthopaedic management. The prognosis was grim, particularly for Type 1 and Type 2 SMA, with significant morbidity and mortality. The unmet need for therapies that could address the underlying genetic defect was substantial.¹

The Therapeutic Revolution and its Consequences

The market for SMA treatment began to change dramatically with the approval of the first disease-modifying therapies.¹ These treatments, including antisense oligonucleotides, gene therapies, and small molecules, work by increasing SMN protein levels.¹ Nusinersen, an antisense oligonucleotide, modulates SMN2 splicing to produce more full-length SMN

protein. Onasemnogene abeparvovec, a gene therapy, delivers a functional copy of the SMN1 gene.¹ Risdiplam, a small molecule, also modifies SMN2 splicing.¹ These therapies have fundamentally altered the disease trajectory, particularly when administered pre-symptomatically or in early infancy.¹

The impact of these therapies has been profound in pediatric populations. Infants treated early have achieved motor milestones, such as sitting and walking, that were previously unattainable for their SMA type. Survival rates have improved significantly, and the need for permanent ventilation has decreased. This success means that a cohort of individuals who would not have survived childhood, or would have lived with severe disability, are now entering adolescence and early adulthood. This demographic shift necessitates a re-evaluation of long-term care strategies.¹

Emerging Challenges in Adult SMA Care

As these treated SMA patients age, clinicians are encountering a new set of challenges that extend beyond the traditional pediatric focus on motor development and acute complications. The long-term effects of chronic SMN deficiency, even with treatment, are not fully understood.¹ While motor function improves, it may not normalize, and patients often continue to experience some degree of muscle weakness and fatigue. The cumulative impact of years of muscle weakness on skeletal health, joint integrity, and overall mobility requires ongoing assessment and management. For a comprehensive understanding of rare diseases and their management, clinicians may find the Oxford Handbook of Clinical Medicine (11th ed) a valuable resource.

Respiratory function remains a critical concern. While early treatment reduces the severity of respiratory compromise, many adult SMA patients may still have reduced lung capacity, weakened cough, and increased susceptibility to respiratory infections. Sleep-disordered breathing is also common and requires careful monitoring. Proactive management, including non-invasive ventilation and airway clearance techniques, is essential to prevent complications and maintain quality of life. This is particularly true for patients who may have experienced periods of respiratory distress in infancy before treatment initiation.

Nutritional status and gastrointestinal issues are also prevalent. Dysphagia, gastroesophageal reflux, and constipation can persist, necessitating dietary modifications, feeding support, and regular gastrointestinal evaluations. Bone health is another significant area of concern. Chronic immobility, corticosteroid use in some patients, and potential metabolic effects of SMA can contribute to osteopenia and osteoporosis, increasing fracture risk. Regular bone density monitoring and interventions to promote bone health are essential for preventing fractures. Clinicians should also be aware of the broader implications of rare diseases, as highlighted in our coverage on rare disease patients facing hurdles beyond diagnosis and treatment.

Multidisciplinary Approach and Psychosocial Support

Managing adult SMA patients effectively requires a truly multidisciplinary approach.¹ This involves neurologists, pulmonologists, gastroenterologists, orthopaedists, physical and occupational therapists, nutritionists, and mental health professionals. Coordination of care across these specialties is paramount to address the complex and interconnected needs of these individuals. The transition from pediatric to adult care services can be particularly challenging, often involving fragmented care and a lack of specialized adult SMA clinics. Establishing clear transition protocols and dedicated adult SMA centers is vital.

Psychosocial well-being is another critical, often overlooked, aspect of care. Growing up with a chronic, progressive

condition, even one that is now treatable, can have significant psychological impacts. Adult SMA patients may face challenges with independence, employment, relationships, and mental health. Providing access to psychological counseling, peer support groups, and vocational rehabilitation services is essential to support their overall well-being and integration into society. The long-term implications of living with a rare genetic disorder extend beyond physical symptoms, as seen in discussions around understanding the genetic basis of rare skin disorders.

The long-term efficacy and safety of the disease-modifying therapies themselves in adult populations are also subjects of ongoing study.¹ While these treatments have shown sustained benefits in pediatric trials, data on decades of use in adults, particularly those who started treatment in infancy, are still accumulating. Questions remain regarding optimal dosing, potential long-term side effects, and the need for continued treatment throughout life.¹ The field needs robust, long-term observational studies and registries to track outcomes in this emerging adult population.¹ This is a common challenge in rare diseases, where long-term data can be scarce, as discussed in the context of transthyretin amyloid cardiomyopathy remaining underdiagnosed.

The Need for Standardized Guidelines and Research

Current clinical guidelines for SMA largely focus on pediatric populations and do not fully address the unique needs of adults who have received early, sustained treatment. There is an urgent need to develop updated, comprehensive guidelines specifically for adult SMA care, incorporating the latest evidence on long-term outcomes, complications, and best practices for multidisciplinary management. These guidelines should cover areas such as respiratory surveillance, bone health, nutritional support, pain management, and psychosocial interventions. The development of such guidelines will help standardize care and ensure that all adult SMA patients receive appropriate and consistent management. Research efforts must also shift to focus on the adult SMA population. This includes studies investigating the natural history of treated SMA into adulthood, identifying biomarkers for disease progression and treatment response in adults, and evaluating the effectiveness of various supportive interventions. Understanding the specific challenges and comorbidities that emerge in adulthood will inform the development of targeted therapies and management strategies. The goal is not just survival, but a high quality of life and maximal functional independence for these individuals. This mirrors the broader push in rare diseases to aim for clinically meaningful endpoints beyond just survival.

The open-label nature of many early SMA trials is an obvious caveat when interpreting long-term outcomes, as is the heterogeneity of patient populations and treatment initiation times. The field must move towards more standardized data collection and outcome measures to allow for meaningful comparisons across studies and patient cohorts. This will be essential for refining treatment strategies and optimizing care for a generation of patients whose lives have been fundamentally altered by therapeutic advances. The journey for these patients is far from over; it has merely entered a new, uncharted phase.

CLINICAL IMPLICATIONS

The success of early SMA therapies has created a new clinical imperative: we must now learn how to care for adults who would not have survived childhood a decade ago. This is not merely an extension of pediatric care; it demands a distinct understanding of chronic disease management in a population with unique needs and evolving complications. General practitioners and specialists alike will increasingly encounter these patients, and a lack of preparedness will lead to suboptimal outcomes.

Clinicians must move beyond a singular focus on motor function. While motor function is a significant aspect, it is only one piece of a complex puzzle. Respiratory, nutritional, skeletal, and psychosocial health require equally rigorous attention. Integrated, multidisciplinary clinics are not a luxury but a necessity for these patients, ensuring coordinated care that addresses the whole person.

The industry, having delivered these life-altering therapies, now has a responsibility to support the development of long-term care infrastructure and research. This includes funding for adult SMA centers, registries to track outcomes, and studies to understand the adult disease phenotype. The initial investment in drug development must be matched by an investment in the lifelong well-being of these patients.

For patients and their families, this new era brings hope but also new uncertainties. They need clear pathways for transitioning from pediatric to adult care, access to specialized services, and robust psychosocial support. The goal is not just to keep them alive, but to enable them to thrive as independent, contributing members of society, a goal that requires a concerted effort from all stakeholders.

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REFERENCES

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