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Editorial

Spinal Muscular Atrophy: A Turning Point in Neuromuscular Medicine

Huseyin Erdal

Department of Medical Genetics, Aksaray University Faculty of Medicine, Aksaray, Türkiye

Spinal muscular atrophy (SMA), once regarded as a relentlessly progressive and untreatable genetic disorder, is now at the forefront of one of the most dramatic transformations in neuromuscular medicine. Characterized by degeneration of motor neurons in the spinal cord, SMA results in progressive muscle weakness, respiratory failure, and in its most severe forms, early mortality. Yet today, hope replaces hopelessness, as recent breakthroughs in molecular medicine have redefined the prognosis and expectations for individuals born with this condition. SMA is most commonly caused by biallelic mutations in the SMN1 gene, leading to deficiency of survival motor neuron (SMN) protein. The presence of SMN2, a gene paralog that produces a small amount of functional SMN protein, plays a pivotal role in determining disease severity. This unique genetic architecture opened the door to targeted therapies that enhance SMN protein production- ushering in a new era of precision medicine.

The development and approval of treatments mark a revolution. These therapies, employing antisense oligonucleotides, gene replacement vectors, or splicing modifiers, offer not just symptomatic relief but disease-modifying potential. Infants who once faced inevitable decline can now sit, stand, and in some cases, walk milestones previously deemed unreachable. However, this progress brings forth new challenges and responsibilities. Early diagnosis is now critical for therapeutic success, particularly as outcomes are dramatically improved when treatment is initiated pre-symptomatically. This has spurred a global push for newborn screening programs, which remain inconsistently implemented despite overwhelming evidence of benefit.

Despite these incredible strides, challenges remain. Access to these highly specialized and expensive treatments is a global issue, demanding equitable healthcare policies and robust support systems. The long-term effects of these therapies are still being studied, and continued research is crucial to optimize dosages, explore combination therapies, and address the residual weakness that some individuals may still experience. Furthermore, the psychosocial support for families navigating a new reality of living with SMA, often after years of despair, is critical.

The story of SMA is a powerful narrative of scientific perseverance, patient advocacy, and the transformative power of biomedical innovation. It reminds us that even in the face of devastating rare diseases, hope is not only possible but can be realized through dedicated research and a collective commitment to improving human health. As we look to the future, the continued progress in SMA treatment stands as a beacon for other rare genetic disorders, inspiring a new era where incurable becomes a temporary descriptor, and the promise of a fuller life becomes a reality for more and more individuals.

With warm regards,

Huseyin Erdal

Editor-in-Chief

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Address for correspondence:
Huseyin Erdal.
Department of Medical Genetics, Aksaray University Faculty of Medicine, Aksaray, Türkiye
E-mail: herdalyfa@gmail.com

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