Gene Therapy in the Era of "Incurable": The Promise and Price of Nusinersen for SMA

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Abstract

The advent of gene therapy offers unprecedented hope for treating genetic disorders like Spinal Muscular Atrophy (SMA), yet its transformative potential is often undermined by prohibitive costs. This paper focuses on Nusinersen, an effective therapy that modifies the SMN2 gene to produce life-sustaining proteins, significantly altering the prognosis for SMA patients. Despite its clinical success in restoring motor functions and stabilizing disease, Nusinersen's exorbitant price tag transforms a medical breakthrough into a financial battleground, restricting access primarily to those with substantial resources or government support. By contrasting the barriers to care with the positive model of public funding in Hong Kong, this work contends that the right to advanced medical treatment should be universal. It concludes that genetic therapies like Nusinersen must be recognized as essential healthcare, guaranteed to all patients regardless of their economic standing.

Introduction

We are living in an era where the word 'incurable' is slowly, painstakingly, being rewritten. With tylenol curing a simple headache, to the development of gene therapy in treating distinct genetic disorders, the scope of what medicine is capable of has expanded immensely. The usage of gene therapy involves introducing genetic material into a patients' cells, aimed to indemnify abnormal genes in order to produce a therapeutic yet necessary protein for daily body functions [1]. As a result, the development of gene therapy has provided the vital option of treatment for patients with genetic disorders once deemed as 'incurable'. However despite the clinical success of the particular treatment, it can impose a significant financial burden on patients and families who have no viable alternatives. This in turn, poses the paradox 'Why has gene therapy turned into a financial battleground due to its increasing effectiveness and precision on treating genetic disorders?'.

Spinal Muscular Atrophy (SMA)

One of the most well known gene therapy is Nusinersen, a direct gene therapy targeting the SMN2 gene in patients diagnosed with Spinal muscular atrophy (SMA) [2]. SMA is a severe genetic neuromuscular disease caused by biallelic mutations and deletions in the SMN1 gene. This results in the inability to produce necessary SMN protein, which is crucial for the health and function of motor neurons – the nerve cells that transmit signals from the spinal cord to the muscle, allowing movements such as walking and breathing. However, because humans obtain SMN2, a nearly identical backup gene of SMN1, it can also produce SMN protein with limitations, as the SMN2 gene falls short of exon 7, the SMN protein produced is often not full length nor functional due to a single nucleotide difference when compared with SMN1 [3].

Limitations of Nusinersen as a Treatment

This is where Nusinersen plays a role in directly targeting the molecular limitations in the SMN2 gene. Nusinersen is an antisense oligonucleotide assigned to bind to the specific intronic

splicing site on the SMN2 pre-messenger RNA, resulting in the inclusion of exon 7, thereby increasing the production of functional full-length SMN protein [4]. First developed in 2004, it is the first approved disease-modifying therapy for SMA, and its clinical success has fundamentally altered the survival outlook for affected patients. Before this treatment, options for SMA were limited due to its rarity, where the most severe consequence often led to infancy and prenatal deaths [5].

Nusinersen benefits suffering patients through its continuous clinical success and effectiveness in restoring SMN proteins for motor functions, as seen through its ability to restore basic motor movements of sitting and walking in infants with Type 1 SMA [6], and to stabilize disease progression in patients diagnosed in late childhood and adulthood, with a young adult with Type 2 SMA testifying to raising her arms to wash her own hair for the first time in years after receiving the therapy [7]. However, while the core of the treatment lies in its effectiveness, it poses practical challenges, particularly for pediatric patients who may require sedation or for those with severe spinal curvature, where accessing the intrathecal space for injection is impractical. Additionally, Nusinersen comes at an extreme cost, from $750,000 for the first year to $375,000 annually thereafter [8]. For patients who can barely afford private care, this treatment is impossible to reach, yet for those with stable income, this price instills financial strain, and with out-of-pocket expenses, it is unrealistic to afford without government funding. What should be a basic human right, becomes a financial war, and far too many families have witnessed the death of loved ones.

Potential Solutions to the Limitation

In a world filled with technology, Nusinersen should be used to treat SMA, where newborn screening allows for early detection and treatment should begin as soon as possible given the disease's rapid progression. Early interference also prevents symptoms from blocking the usage of gene therapy where treatment becomes impractical. Though high costs remain a barrier for many families, in Hong Kong, where I live, the government has designated this therapy as a publicly funded drug, making it accessible to all, including those around me, making this an extremely effective and available treatment [9].

With the rapid development of genetic therapies today, access to treatment should not be determined by financial capability. It should be a basic human right, ensuring that no patient faces additional strain after a genetic disorder diagnosis. While public funding in my city protects citizens from the financial burden of affording nusinersen, the right to such treatment should be universally guaranteed, granting equal access for all patients.

Conclusion

With the rapid development of genetic therapies today, access to treatment should not be determined by financial capability. It should be a basic human right, ensuring that no patient faces additional strain after a genetic disorder diagnosis. While public funding in my city protects citizens from the financial burden of affording nusinersen, the right to such treatment should be universally guaranteed, granting equal access for all patients.

Work Cited

[1]: Center for Biologics Evaluation and Research, “What is gene therapy?,” U.S. Food and Drug Administration, https://www.fda.gov/vaccines-blood-biologics/cellular-gene-therapy-products/what-gene-therapy (accessed Mar. 5, 2026).

[2]: “Spinal muscular atrophy (SMA) - diseases,” Muscular Dystrophy Association, https://www.mda.org/disease/spinal-muscular-atrophy (accessed Mar. 5, 2026).

[3]: M. C. A. Virata, R. M. R. Abello, E. M. B. Brillante, and M. B. Corneta, “SMN2 as a therapeutic target in spinal muscular atrophy: Advances in gene expression modulation,” Journal of Translational Genetics and Genomics, https://www.oaepublish.com/articles/jtgg.2025.56 (accessed Mar. 5, 2026).

[4]: J. Qiu et al., “History of development of the life-saving drug ‘Nusinersen’ in spinal muscular atrophy,” Frontiers in cellular neuroscience, https://pmc.ncbi.nlm.nih.gov/articles/PMC9414009/ (accessed Mar. 5, 2026).

[5]: D. Lopes, “Gene therapy for spinal muscular atrophy effective as first- or second-line therapy,” The Medical Xchange, https://themedicalxchange.com/en/2021/06/30/2434_gene-therapy-for-spinal-muscular-atrophy-effective-as-first-or-second-line-therapy/ (accessed Mar. 5, 2026).

[6]: Briefing document to the clinical trial readiness in spinal muscular, https://www.ema.europa.eu/en/documents/other/briefing-document-clinical-trial-readiness-spinal-muscular-atrophy-sma-sma-europe-treat-nmd-and-european-medicines-agency-meeting_en.pdf (accessed Mar. 4, 2026).

[7]: Spring 2013 issue 15 families of spinal muscular atrophy www.curesma.org, https://www.curesma.org/wp-content/uploads/2019/10/Directions_Spring2013.pdf (accessed Mar. 4, 2026).

[8]: M. Haché et al., “Intrathecal injections in children with spinal muscular atrophy: Nusinersen clinical trial experience,” Journal of child neurology, https://pmc.ncbi.nlm.nih.gov/articles/PMC4871174/ (accessed Mar. 5, 2026).

[9]: “Rare disease drug set for HK,” Hong Kong’s Information Services Department, https://www.news.gov.hk/eng/2018/03/20180301/20180301_152631_917.html (accessed Mar. 5, 2026).