INDIAN FEDERATION OF NEUROREHABILITATION

Journal of Federation of Neurorehabilitation
2025, Vol. 2, No. 2, pp. 12–17
DOI: 10.54646/IFNR.2025.13
jifnr.ifnr.org

CASE REPORT

Reviving mobility: the impact of activity-based rehabilitation on motor function in a child with SMA type II after gene therapy – a case report

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Received: 08 December 2025; **Accepted:** 22 December 2025; **Published:** 16 February 2026

Background: Spinal Muscular Atrophy (SMA) Type II is a genetic neuromuscular disorder causing progressive muscle weakness. Gene therapy with Onasemnogene Abeparvovec (Zolgensma) has improved outcomes when combined with early rehabilitation.

Objective: To assess the impact of activity-based physiotherapy on motor function, postural control, and quality of life in a child with SMA Type II post-gene therapy.

Methods: A 1-year-11-month-old female received gene therapy at 18 months, followed by a 12-month multidisciplinary physiotherapy program including postural alignment, functional sitting training, neuromuscular stimulation, standing frame, respiratory exercises, and aquatic therapy. Outcome measures included Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND), Hammersmith Functional Motor Scale Expanded (HFMSE), PedsQL, and manual muscle testing.

Results: CHOP INTEND improved from 54 to 64, HFMSE from 29 to 35, and PedsQL from 40 to 34. Notable improvements were observed in trunk control, dynamic sitting, and functional transitions.

Conclusion: Early, structured rehabilitation post-gene therapy can enhance motor outcomes and quality of life in children with SMA Type II.

Keywords: activity-based physiotherapy, motor function, postural control, quality of life, SMA type II, neuromuscular stimulation, aquatic therapy

Introduction

Spinal Muscular Atrophy (SMA) is a rare autosomal recessive neuromuscular disorder caused by deletion or mutation of the Survival Motor Neuron 1 (SMN1) gene. This leads to degeneration of alpha motor neurons, resulting in progressive muscle weakness and atrophy (1). SMA is classified into four types, with Type II (Dubowitz disease) presenting between 6 and 18 months. Affected children can sit independently but never achieve unaided walking (2).

Onasemnogene Abeparvovec, an adeno-associated viral vector-based gene therapy, has emerged as a transformative

treatment for SMA. Additional pharmacologic therapies include nusinersen and risdiplam, which modulate SMN2 gene splicing. Early therapeutic intervention is crucial to preserve motor neurons and optimize outcomes (3, 4).

This report (5) highlights the role of early, activity-based physiotherapy post-gene therapy in improving motor function, postural control, and quality of life in a child with SMA Type II.

Case presentation

A 1-year-11-month-old female presented to the Neuromuscular Centre of Excellence, Department of Neurology, at Christian Medical College and Hospital with a diagnosis of SMA Type II. Born full-term via lower segment cesarean section (LSCS) with normal birth parameters and early neonatal health, she received phototherapy for two days postnatal. At the time of presentation, the child's anthropometric measurements were as follows: weight 10.1 kg, height 79 cm, and head circumference 46 cm. She showed delayed motor milestones: neck control at 6 months and rolling at 10 months, but an inability to transition independently or sit without support.

Investigations

Multiplex ligation-dependent probe amplification (MLPA) confirmed homozygous deletion of SMN1 exons 7 and 8 and heterozygous duplication of SMN2 exons 7 and 8. Nerve conduction velocity (NCV) revealed reduced amplitude in the left tibial nerve.

Medical management

At 1 year and 6 months of age, she received gene therapy (Zolgensma) from the USA and has since been on syrup Risdiplam 2.5 ml OD as part of her ongoing treatment.

Rehabilitation protocol

Initial examination

Comprehensive physiotherapy assessments were conducted at baseline, after 6 months, and after 12 months of intervention. Assessments included:

- **Motor Function:** Evaluated using the Hammersmith Functional Motor Scale Expanded (HFMSE) and Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND) (**Figure 1**; **Table 1**).
- **Quality of Life:** Assessed with the PedsQL^{TM} 3.0 Neuromuscular Module.
- **Muscle Tone and Strength:** Documented through manual muscle testing and goniometric range of motion measurements (**Table 2**).

At baseline, the child demonstrated generalized hypotonia, delayed motor milestones, and significant weakness in both proximal and distal musculatures.

The primary objectives were to:

- Improve trunk control and dynamic sitting balance.
- Facilitate functional transitions, such as sit-to-stand.
- Promote symmetrical weight-bearing and enhance postural endurance.
- Encourage spinal alignment and controlled reaching.
- Enhance respiratory function and breath control.

Secondary goals included reducing compensatory strategies and improving quality of life.

A 12-month intensive physiotherapy program was initiated, consisting of the following components:

Postural alignment

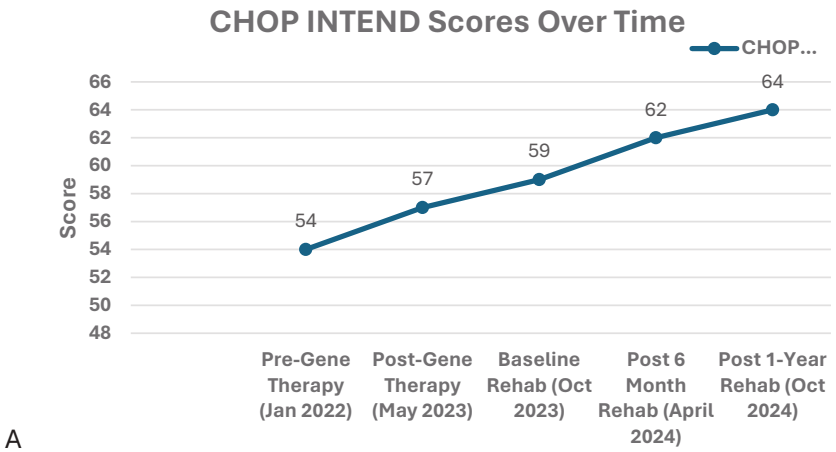
Primary care in the initial phase emphasized 24/7 alignment and postural management through various supported postures. Interventions such as night splints, serial casting, and pelvic alignment with 30° hip abduction were implemented to prevent deformities like coxa valga. Early postural care is crucial in pediatric populations to support musculoskeletal development during growth. Proper alignment, frequent position changes, and supported sitting help maintain symmetry, prevent pelvic obliquity and scoliosis, and reduce secondary complications. Consistent early intervention ensures joint integrity and promotes stable posture as the child grows.

TheraTogs orthotic garment

- Worn 4–6 hours/day, 7 days/week
- Primarily during active therapy and upright play. The application focused on trunk and pelvic alignment, enhancing core activation and reducing postural collapse during sitting, standing, and transitional tasks.
- TheraTogs facilitated symmetrical engagement, improved alignment, and promoted optimal motor patterns, especially in dynamic sitting and pre-standing tasks (**Figures 2B, F**).

Targeted training

- Frequency: 5 days/week, 20–25 minutes/session



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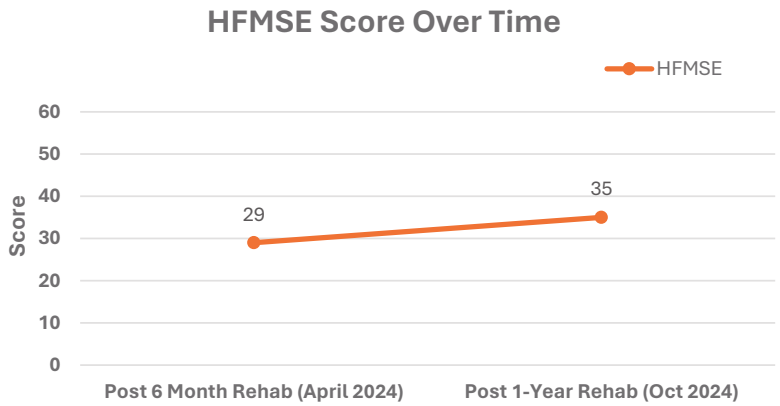

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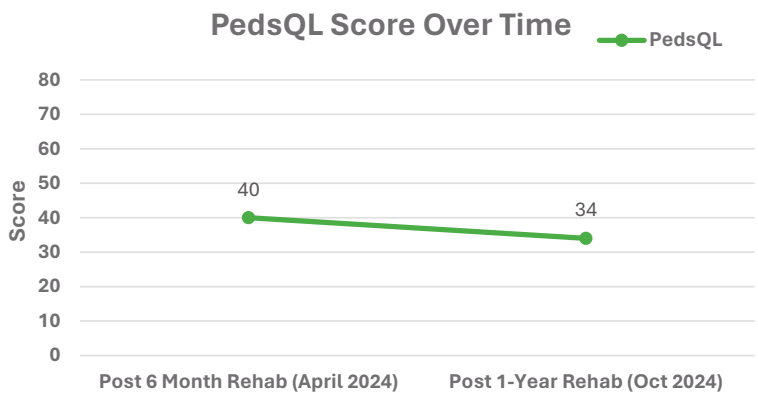

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FIGURE 1 | (A) Graph of CHOP INTEND, (B) Graph of HFMSE scores, and (C) PEDQOL scores across rehabilitation timeline.

- Tasks included upward and downward (frontal plane) reach-outs and targeted muscle activation (anterior pushing activity for serratus anterior and downward pressure techniques for latissimus dorsi) for scapular stability. Interventions included pediatric alignment training with point-specific degrees of freedom and strengthening activities to support postural control and upper limb function (**Figures 2A, E**).

Neuromuscular electrical stimulation (NMES)

- Frequency: 3 days/week, 15–20 minutes
- Spinal muscle to initiate baseline trunk activation and support core stability, gluteal with 25 repetitions during sit-to-stand transitions to improve lower

TABLE 1 | Pre and post intervention scores of Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND), Hammersmith Functional Motor Scale Expanded (HFMSE), PedsQL.

Assessment timepoint	CHOP INTEND	HFMSE	PedsQL
Pre-Gene Therapy (Jan 2022)	54/64	-	-
Post-Gene Therapy (May 2023)	57/64	-	-
Baseline Rehab (Oct 2023)	59/64	-	-
Post 6 Month Rehab (April 2024)	62/64	29/66	40/84
Post 1-Year Rehab (Oct 2024)	64/64	35/66	34/84

TABLE 2 | Pre and post intervention grades of manual muscle testing.

Muscle group	Post 6 months (right/left)	Post 12 months (right/left)
Hip		
Hip Flexors	Grade 2-/2-	Grade 3/3
Hip Extensors	Grade 3-/3-	Grade 3/3
Hip Abductors	Grade 2-/2-	Grade 3-/3-
Hip Adductors	Grade 2-/2-	Grade 3-/3-
Internal Rotators	Grade 1/1	Grade 2/2
External Rotators	Grade 1/1	Grade 2/2
Knee Flexors	Grade 3-/3-	Grade 4/4
Knee Extensors	Grade 3-/3-	Grade 4/4
Ankle Planter Flexors	Grade 2/2	Grade 3/3
Ankle Dorsiflexors	Grade 2/2	Grade 3/3
Ankle Invertors	Grade 2/2	Grade 3/3
Ankle Evertors	Grade 2/2	Grade 3/3

limb strength and facilitate functional movement, and adductor stimulation integrated with functional transitions and weight-shift exercises (**Figure 2H**).

Standing frame

- Duration: 30–45 minutes, 2 days/week
- As part of the postural management approach, proper alignment was maintained using lateral structural support for the trunk and vertical positioning of the lower limbs.
- Activities included single-leg stance, cross-body reaches, and scapular and trunk engagement exercises (**Figure 2B**).

Breathing exercises

- Frequency: 5x/week, 10–15 minutes
- Techniques included balloon blowing, Flower and Candle Breathing, which combined slow, controlled inhalation (smelling the flower) with gentle, prolonged exhalation (blowing out the candle) to encourage diaphragmatic breathing, straw sipping, and paper blowing to enhance respiratory endurance.

Aquatic therapy (Halliwick method)

- Frequency: done for 3 days for 45 minutes and was then recommended to continue at home
- Conducted in a 7.5 × 25 m pool at 30–32°C, the parent was present during the therapy sessions to enhance the child's motivation and minimize his fear of water. Therapy consists of 5–10 minutes of warm-up (such as breathing and walking [with the help of elevation]) and stretching exercises, 40 minutes of the Halliwick concept (lying in different directions, rolling, and swimming), and 5–10 minutes of cool-down exercises (walking) (**Figure 2G**).

Sensorimotor priming

- Frequency: 5 days/week
- Duration: 10–15 minutes/session
- Techniques included passive oscillations, proprioceptive stimulation with vibration and tapping, spinal mobilizations, and oblique muscle activation using pressure-release techniques (**Figures 2C, D**).

Result

Following gene therapy with **Onasemnogene Abeparvovec** and the initiation of a structured physiotherapy program, the child demonstrated significant improvements in motor function and postural control over a 12-month period.

Discussion

This case study illustrates the significant functional gains in a child with SMA Type II following early genetic intervention with Onasemnogene Abeparvovec (Zolgensma) and a comprehensive, structured activity-based physiotherapy program. The integration of intensive motor training, neuromuscular stimulation, and aquatic therapy promoted improvements not only in isolated muscle groups but also in coordinated functional tasks such as dynamic sitting, transitions, and early weight-bearing.

The increase in the CHOP INTEND score from 54/64 pre-gene therapy to a full 64/64 score after one year highlights the synergistic role of gene therapy and rehabilitation. While Zolgensma addresses the genetic root cause by restoring SMN1 expression, physiotherapy supports neuroplasticity, postural control, and motor relearning—particularly vital

FIGURE 2 | Rehabilitation. (A, B) Pre- and post-intervention standing posture. (C, D) Pre- and post-intervention quadripod position. (E, F) Pre- and post-sitting posture with and without TheraTogs. (G) Aquatic therapy. (H) neuromuscular electrical stimulation (NMES) on spinal muscles.

for children with existing motor delay or muscle imbalance (5, 6).

A HFMSE score of 35/66, while below full functional independence, still represents a meaningful improvement in gross motor capabilities, especially considering the initial clinical presentation of generalized hypotonia and inability to sit independently. This score places the child in a moderate function range, and continued therapy can further facilitate milestone achievement (7).

Notably, the use of aquatic therapy provided a low-gravity environment that enabled safe practice of transitions and upright postures, reducing compensatory movements and promoting symmetrical engagement of trunk and limb musculature. Neuromuscular electrical stimulation (NMES) was applied with functional intent (e.g., during sit-to-stand training), which may have accelerated muscle recruitment and improved postural endurance (8, 9).

Importantly, early intervention is emphasized. Initiating both gene therapy and rehabilitation at a young age capitalizes on the child's neurodevelopmental window, allowing for better outcomes compared to delayed therapy. This aligns with current literature advocating

for early genetic screening and treatment initiation to prevent irreversible motor neuron loss. Limitations of this case report include its single-subject design and absence of a control comparison, warranting future studies with larger cohorts (10).

Conclusion

This case highlights the critical role of early, intensive, activity-based rehabilitation in maximizing functional outcomes in children with SMA Type II following gene therapy. A structured 12-month physiotherapy program, incorporating neuromuscular stimulation, functional training, and aquatic therapy, led to measurable improvements in motor function, postural control, and quality of life.

These findings emphasize the importance of integrating multidisciplinary rehabilitation protocols immediately following genetic intervention to fully harness the therapeutic potential of gene therapy. Future research involving larger sample sizes and longer-term follow-up is necessary to establish evidence-based rehabilitation guidelines for SMA.

Ethics statement

The patient's mother has given consent for publication and photographs.

Funding

No Funding was received for the study.

Author contributions

MCK: Conceptualization, Data curation, Supervision, Writing – original draft, Writing – review & editing. MP: Conceptualization, Writing – review & editing. VJ: Investigation, Writing – review & editing. MS: Investigation, Writing – review & editing. AB: Conceptualization, Data curation, Writing – original draft, Writing – review & editing.

Acknowledgments

We thank the patient for permitting us to include his data in this article.

Conflict of interest

The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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