Case Report

DOI: 10.7860/JCDR/2026/81639.22695

Physiotherapy Section

New Horizons in Physiotherapy for Spinal Muscular Atrophy (SMA) Type 1 in the Era of Disease-modifying Treatments: A Case Report

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ABSTRACT

Spinal Muscular Atrophy (SMA) type 1 is a severe autosomal recessive neuromuscular disorder caused by SMN1 gene mutations, leading to progressive muscle weakness and respiratory compromise. Without intervention, affected infants can rarely sit independently or live beyond two years of age. The advent of disease-modifying therapies and the timely implementation of physiotherapy have changed the disease trajectory and helped in increased survival. This report describes a two-year and five-month-old female child with genetically confirmed SMA type 1 on disease modifying treatment and regular physiotherapy who achieved independent sitting - an uncommon milestone for this phenotype. During hospitalisation for bronchopneumonia at a tertiary care hospital, physiotherapy was continued with chest physiotherapy and truncal activation exercises, progressing to functional strengthening and postural control activities. Functional gains were documented using the Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP-INTEND) and the Revised Hammersmith Scale for Spinal Muscular Atrophy (RHS-SMA). This case underscores the vital role of structured physiotherapy in translating pharmacological advances into meaningful functional recovery in children with SMA type 1.

Keywords: Early intervention, Motor neuron disease, Paediatric rehabilitation

CASE REPORT

A two-year and five-month-old female child with a known diagnosis of SMA Type 1 (confirmed at five months of age) was admitted to a tertiary care hospital with complaints of cough, cold, and fever persisting for three days. On evaluation, she was diagnosed with bronchopneumonia with respiratory distress and referred for physiotherapy to aid airway clearance, improve lung expansion, and maintain functional abilities during hospitalisation.

At the time of physiotherapy referral, the child presented with retained secretions and breathing difficulty, a forward-bent posture in sitting with inability to attain sitting independently, reduced spontaneous limb movements (especially in lower limbs) and inability to stand without support.

This was the first child born to non-consanguineous parents, with no known family history of neuromuscular disorders, genetic conditions, or psychosocial stressors. The antenatal period was significant for intrauterine growth retardation detected during the third trimester. The child was delivered at term via normal vaginal delivery with an episiotomy and had a birth weight of 3.1 kg. The baby cried immediately after birth, though the cry was noted to be weak. The APGAR score at one and five minutes was 9/10.

A chronological summary of the child's medical and rehabilitation history was documented to highlight key diagnostic and therapeutic milestones. The major clinical events and interventions are outlined in [Table/Fig-1] [1-4].

Developmental and functional milestones were evaluated to understand the child's baseline motor abilities, indicating a developmental age of eight months (sitting without support) in the gross motor domain and two years (making a tower of 6 blocks) in the fine motor domain.

The child was well aroused and alert, with a normal affect and good attention; vision, speech, and hearing were intact, while motor activity was noted to be reduced in the lower limbs.

Age	Event/Observation
5 months	Genetic testing showed homozygous deletion of exons 7 and 8 in the SMN1 gene (SMN1:SMN2 ratio = 0:2). Child diagnosed with SMA type 1
7 months	Risdiplam initiated at tertiary care hospital; physiotherapy started, CHOP INTEND† [1] score: 17/64
1 year	HINE‡ [2] score of 51/78, CHOP INTEND score of 45/64. Risdiplam and home physiotherapy were continued. The child was using Taylor's brace for the prevention of scoliosis development.
2 years 1 month	Diagnosed and hospitalised with bronchopneumonia, on non-invasive ventilation for 3 days, discharged within 6 days. Risdiplam and home physiotherapy continued.
2 years 2 months	Follow-up assessment documented CHOP-INTEND score of 51/64 and RHS-SMA§ [3] score of 8/69.
2 years 5 months	Diagnosed and hospitalised with bronchopneumonia and respiratory distress, on oxygen supplementation with nasal prongs, Risdiplam ongoing, in hospital physiotherapy was started.
Status at the time of physiotherapy referral	Sitting with support, grade 4 dyspnoea on modified Medical Research Council Dyspnoea scale [4], on oxygen supplementation with an oxygen mask, active mobility in gravity eliminated plane, oral feeding with no swallowing difficulties. Chest physiotherapy and neuro physiotherapy started.

[Table/Fig-1]: Timeline of events [1-4].
†CHOP INTEND: Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders; ‡HINE: The Hammersmith Infant Neurological Examination §RHS-SMA: Revised Hammersmith Scale for Spinal Muscular Atrophy

Respiratory examination revealed reduced chest expansion at the inframammary level, while excursion was symmetrical. On auscultation, air entry was reduced bilaterally in the lower zones with coarse crepitations.

Deep tendon reflexes were hyporeflexive in the bilateral upper limbs and absent in the lower limbs. Hypermobility and increased range of motion were observed across all joints with generalised hypotonia in all four limbs. Manual Muscle Testing (MMT) revealed predominant proximal weakness with relatively preserved distal control in the

upper limbs [5]. Trunk MMT showed poor strength, and lower limb MMT demonstrated significant weakness in hip and knee extensors [Table/Fig-2].

Muscle group	Left	Right
Shoulder flexors	3	2+
Shoulder abductors	3	3-
Elbow flexors	3	3
Elbow extensors	3	3
Wrist flexors	3	3
Wrist extensors	3	3
Hip flexors	2-	2
Hip extensors	2-	2-
Hip abductors	2	2
Knee flexors	2	2
Knee extensors	2+	2+
Ankle dorsiflexors	2	2
Ankle plantar flexors	2+	2+
Abdominals	1	
Back extensors	1	

[Table/Fig-2]: Manual Muscle Testing (MMT).

Postural assessment [Table/Fig-3] revealed forward head posture with increased thoracic kyphosis.

Lateral view	Anterior view	Posterior view
Forward head posture Increased cervical lordosis Protracted shoulders Increased thoracic kyphosis (C-shaped spine) Lack of lumbar lordosis Posterior pelvic tilt	Right shoulder elevated Head and trunk lean towards the left	Increased left neck shoulder angle Left shoulder and scapula depressed Lateral convexity to the right in thoracic region Lateral convexity to the left in lumbar region

[Table/Fig-3]: Postural assessment.

Functional assessment was conducted using both the CHOP-INTEND and the RHS-SMA [1,3]. Both of these are freely accessible clinical outcome measures for children with SMA. The RHS, although primarily validated for SMA types 2 and 3, was used to evaluate gross motor and postural transitions. Items not achievable due to the patient's limited motor function were scored according to the standard RHS protocol, acknowledging that some items may result in floor effects. The CHOP INTEND, a scale specifically validated for SMA Type 1 infants, was employed to capture 16 items related to spontaneous movement, muscle strength, and antigravity control. The combined use of both scales allowed a comprehensive evaluation across the spectrum of motor function, while addressing the limitations of each individual scale.

As per records, scores assessed three months prior were 51/64 on CHOP-INTEND and 8/69 on RHS-SMA. During the current hospitalisation, the scores were 45/64 and 7/69, respectively. The observed reduction reflected the natural course of the disease, compounded by the acute episode of bronchopneumonia.

Informed consent was obtained from the parents for the use of the data obtained from the child for educational and research purposes.

Therapeutic intervention: Physiotherapy during the hospital stay focused on addressing key problems identified on assessment, including reduced chest expansion with retained secretions, generalised hypotonia, impaired postural control, and poor proximal stability. The goals were to improve lung expansion and airway clearance, facilitate tone normalisation, enhance postural and trunk stability, and promote functional mobility. Sessions emphasised neuromotor facilitation for trunk control, proprioceptive and weight-bearing activities for tone regulation, and assisted mobility training such as supported standing. In-hospital physiotherapy was delivered twice daily for 10 days, with every session lasting approximately 30-40 minutes. The intervention protocol was designed in accordance with standard paediatric rehabilitation and neurodevelopmental principles and was tailored to the child's endurance and fatigue level [6,7].

[Table/Fig-4] [6,7] summarises the detailed physiotherapy programme implemented during the hospital stay, outlining therapeutic goals and interventions.

Problem list	Physiotherapy goals	Interventions	Frequency and duration
Reduced chest expansion, diminished air entry, presence of secretions	To improve lung expansion and ventilation, promote effective airway clearance, and optimise overall respiratory function	Airway clearance techniques (Gentle percussion, vibration, and postural drainage) Breathing exercises: Inspiratory exercises: Diaphragmatic breathing, drinking from a straw Expiratory exercises: Glove blowing, Whistle blowing, Making bubbles, incentive spirometer, paper blowing	10 repetitions, 2 sets were given for breathing exercises.
Hypotonia causing impaired movement and postural control	To normalise muscle tone	Tapping: Light, rhythmic tapping Light touch & fast brushing: Sensory stimulation to facilitate muscle tone regulation and improve tactile awareness. Joint compressions: Heavy compressive forces applied to joints to improve proprioception and promote muscle co-contraction for stability.	Tapping: Applied with the fingertips 3-5 times; given in conjunction with other treatment techniques Fast brushing: Applied it over the dermatomes for 3 to 5 seconds and repeated after 30 seconds× 5 reps [6]
Impaired ability to sustain a posture and maintain muscle activation	To promote the sustenance of various positions like kneeling, half kneeling, and sitting	To maintain a supported sitting position with back extensor facilitation using Neuro Developmental Techniques (NDT) To facilitate kneeling with bolster support using NDT	Given for 15-20 minutes, progressed gradually as per facilitation response and fatigue levels [7].
Poor proximal stability affecting upper and lower limb function	To facilitate supported sitting activities combined with upper and lower limb functional strengthening exercises	Reach overhead to get a toy while maintaining upright posture (to activate trunk extensors and facilitate upper extremity overhead movements while maintaining upright posture) Bilateral reaching for toys placed at shoulder/head level to activate deltoids and scapular stabilisers. Push-pull games (Pushing a therapy ball forward) to enhance dynamic trunk control and upper limb strength. Ball kicking games to activate the quadriceps and hip flexors.	10 repetitions each bilaterally × 2 sets
Poor trunk stability and control during limb movements.	To train dynamic trunk control	Reaching while sitting, ball toss, and playing with toys placed above shoulder level to encourage active upper limb engagement and trunk control. Trunk rotations aimed at increasing multidirectional dynamic control.	10 repetitions × 2 sets

Impaired ability to transition from one position to another	To train transitions from side-lying to prone and from supine to sitting	NDT facilitated transition from side-lying to prone to encourage trunk rotation, oblique activation, and movement sequencing. Guided transition from supine → side-lying → sitting using NDT facilitation.	Given for 15-20 minutes, progressed gradually as per facilitation response and fatigue level [7]
Decreased bone mineral density, decreased joint loading and proprioceptive input	To facilitate supported standing to improve bone mineral density	Use standing frames with the hip in slight abduction	10-15 min/ day, once daily.
Decreased abdominal and back extensor strength	To improve core strength and activation of the core during various transitions and postural activities	Pelvic tilts Partial curl-ups (with assistance) Obliques with reaching for a toy (with assistance) Prone on forearms with neck and back extension	10 repetitions × 1 set

[Table/Fig-4]: Therapeutic intervention [6,7].
(The physiotherapy protocol was customised and progressively adjusted to the child's individual requirements, with adequate rest breaks incorporated, and all activities were not administered in a single session and spaced according to her endurance and fatigue levels)

After 10 days of hospitalisation, at the time of discharge, the child was sitting without support under supervision, had grade 2 dyspnoea and was breathing on room air, demonstrated active mobility in gravity-eliminated planes with minimal resistance, and was feeding orally without swallowing difficulties. The mother was educated about the home programme and the importance of regular follow-up.

Later, the home-based physiotherapy was continued for one month under caregiver supervision, with periodic teleconsultations with the hospital physiotherapy team, to review progress and guide exercise progression. This ensured continuity of neurorehabilitation and reinforcement of therapy techniques beyond the hospital setting.

Beyond this period, the family continued the home programme independently, and no further follow-up data were available at the time of reporting.

At one-month follow-up, with ongoing medical and physiotherapy management, the child's condition remained stable, with CHOP-INTEND and RHS-SMA scores of 45/64 and 7/69, respectively, suggesting preservation of functional abilities and prevention of further decline.

Although SMA type 1 is a progressive disorder, the stability of functional scores highlights the role of physiotherapy in promoting recovery following acute illness episodes and preventing further deterioration.

DISCUSSION

Physiotherapy plays a pivotal role in optimising motor outcomes and functional independence in children with SMA type 1, particularly in the era of disease-modifying therapies that have improved survival and motor potential. In the present case, continued individualised neurorehabilitation focusing on respiratory efficiency, proximal stability, and motor facilitation led to measurable improvements in trunk control and functional abilities. Early initiation of airway clearance techniques, including postural drainage, percussion, and breathing exercises, helped maintain lung compliance and prevent atelectasis, which is a common concern in SMA due to weak respiratory musculature. These findings align with the recommendations of Mercuri E et al., who emphasise that consistent chest physiotherapy and upright positioning significantly improve ventilation-perfusion balance and reduce respiratory complications in SMA type 1 [8].

The structured motor programme based on neurodevelopmental principles promoted improved postural, truncal activation, and dynamic postural control through graded sitting and functional reaching tasks. Such interventions encourage cortical reorganisation and neuromuscular adaptation, which are crucial for translating neural preservation into active movement. Similar findings have been reported by Yi YG et al., who demonstrated that early, task-specific physiotherapy can enhance antigravity control and sitting balance even in severe SMA phenotypes [9].

Progressive strengthening through play-based activities such as reaching, pushing, and supported standing improved co-contraction and weight-bearing. Studies by Shin HI and Foead AI et al., support these outcomes, reporting that structured physiotherapy programs emphasising endurance, postural control, and sensory facilitation significantly improve functional performance in SMA children under active rehabilitation [10,11].

In this case, maintenance of CHOP-INTEND and RHS-SMA scores despite disease progression reflects stabilisation of function through prompt physiotherapy intervention along with medical care. These findings reinforce that physiotherapy serves as a critical contributor to motor preservation, translating pharmacological benefits into meaningful functional outcomes. There remains a dearth of literature describing physiotherapy-specific programmes in SMA Type 1; however, limited observational studies and consensus papers suggest that early, individualised neurorehabilitation focusing on respiratory care, proximal stability and task-specific practice can maximise functional preservation and permit translation of pharmacological gains into motor improvements [8-13]. Our case adds evidence that integrated medical and rehabilitative approaches may allow achievement of milestones otherwise rare in this phenotype and it supports the growing consensus that physiotherapy is not merely adjunctive but an essential determinant of motor recovery in SMA Type 1.

CONCLUSION(S)

This case demonstrates that, along with disease-modifying drugs, a structured, goal-directed physiotherapy program can maintain motor function and prevent regression in a child with SMA type 1. Early initiation and continuity of physiotherapy focusing on respiratory care, proximal muscle activation, and postural control led to improvement in trunk stability, sitting balance, and endurance. The child demonstrated stabilisation of functional abilities and achievement of independent sitting, which is a milestone rarely observed in this phenotype.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Jul 07, 2025
- Manual Googling: Dec 18, 2025
- iThenticate Software: Dec 20, 2025 (3%)

ETYMOLOGY: Author Origin

EMENDATIONS: 7

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Jul 04, 2025**
Date of Peer Review: **Oct 25, 2025**
Date of Acceptance: **Dec 22, 2025**
Date of Publishing: **Apr 01, 2026**