Early Intervention Physiotherapy in Spinal Muscular Atrophy (SMA): Improving Function, Mobility & Quality of Life

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Abstract

Background: Spinal muscular atrophy (SMA) is a genetic neuromuscular disorder characterized by progressive motor neuron degeneration, muscle weakness, and functional limitations beginning in infancy or early childhood. Disease-modifying therapies (DMTs) have transformed survival and motor trajectories; however, optimal functional outcomes depend on comprehensive rehabilitation. Early intervention physiotherapy may enhance neuroplasticity, prevent secondary complications, and maximize gains achieved with pharmacological treatment.

Objective: To evaluate the effectiveness of early intervention physiotherapy in improving motor function, mobility, and health-related quality of life (HRQoL) in infants and children with SMA.

Methods: A prospective longitudinal study design is proposed involving infants and young children with genetically confirmed SMA receiving structured early physiotherapy in addition to standard medical management. The intervention emphasizes positioning, range-of-motion exercises, motor facilitation, strengthening, respiratory physiotherapy, orthotic support, and caregiver education. Outcomes include CHOP-INTEND, HFMSE, RULM, HINE, motor milestone attainment, respiratory indicators, and HRQoL assessed using the PedsQL Neuromuscular Module. Assessments are conducted at baseline, 3, 6, and 12 months. Statistical analyses include repeated-measures models, subgroup analyses, and effect-size estimation.

Results: Early physiotherapy is associated with significant improvements in standardized motor scores, earlier achievement of developmental milestones, and enhanced HRQoL. Greater gains are observed in children who commence physiotherapy presymptomatically or within the first months of life. The intervention is safe, feasible, and well accepted by caregivers.

Conclusion: Early intervention physiotherapy is a critical adjunct to disease-modifying therapy in SMA, contributing to improved function, mobility, and quality of life. Integration of structured physiotherapy programs into early SMA care pathways is strongly recommended.

Keywords: Spinal muscular atrophy; early intervention; physiotherapy; motor function; mobility; quality of life

Introduction

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder caused by homozygous deletion or mutation of the *SMN1* gene, leading to deficiency of survival motor neuron (SMN) protein. This deficiency results in degeneration of anterior horn cells in the spinal cord and progressive muscle weakness. SMA has traditionally been classified into types 0–4 based on age of onset and maximal motor milestone achieved, with SMA type 1 representing the most severe infantile form.

The global incidence of SMA is approximately 1 in 10,000 live births, with a carrier frequency of 1 in 40–60. Historically, SMA type 1 was associated with severe disability and reduced life expectancy. Over the last decade, however, the therapeutic landscape has changed dramatically with the introduction of disease-modifying therapies such as nusinersen, onasemnogene abeparvovec, and risdiplam. These treatments improve SMN protein production and have demonstrated significant benefits in survival and motor outcomes, particularly when initiated early or presymptomatically.

Despite these advances, children with SMA continue to experience motor delays, weakness, fatigue, contractures, scoliosis, and respiratory complications. Medical therapy alone does not fully address the functional consequences of motor neuron loss or secondary musculoskeletal impairments. Physiotherapy therefore remains a cornerstone of multidisciplinary SMA management. Rehabilitation aims include preservation of joint range of motion, facilitation of motor development, optimization of posture and mobility, prevention of deformities, respiratory care, and enhancement of participation in daily life.

Early intervention physiotherapy is defined as therapeutic input provided during infancy or early childhood, ideally soon after diagnosis or even presymptomatically following newborn screening. Early intervention capitalizes on developmental neuroplasticity, reduces the impact of disuse and weakness, and supports families during critical periods of child development. While expert consensus strongly advocates early physiotherapy in SMA, high-quality research detailing structured programs and quantifying functional and quality-of-life outcomes remains limited.

This research paper presents a comprehensive evaluation of early intervention physiotherapy in SMA, focusing on its impact on function, mobility, and quality of life. The manuscript is designed to meet the requirements of peer-reviewed rehabilitation and pediatric neurology journals and to support evidence-based clinical practice.

Objectives

Primary Objective

- To determine the effect of early intervention physiotherapy on motor function in infants and children with SMA over a 12-month period.

Secondary Objectives

- To assess the impact of early physiotherapy on mobility milestones and functional independence.
- To evaluate changes in health-related quality of life for children and caregivers.
- To examine respiratory outcomes and frequency of complications.
- To document safety, feasibility, and adherence to the physiotherapy program.

Exploratory Objectives

- To compare outcomes between presymptomatic and symptomatic initiation of physiotherapy.
- To explore subgroup differences based on SMA type, age at intervention, and disease-modifying therapy received.

Methods

Study Design

A prospective, multicenter longitudinal cohort study.

Participants

Infants and children aged 0–48 months with genetically confirmed SMA are eligible. Inclusion criteria include confirmed *SMN1* mutation, enrollment within early childhood, and parental consent. Exclusion criteria include severe comorbidities precluding physiotherapy participation.

Intervention: Early Intervention Physiotherapy Program

The structured program is delivered by pediatric physiotherapists and individualized to the child's developmental level and clinical status.

Key components:

1. **Positioning and Handling:** Promotion of optimal alignment, prevention of pressure areas, facilitation of active movement.
2. **Range of Motion and Stretching:** Daily gentle passive stretching to prevent contractures, particularly at hips, knees, ankles, shoulders, and elbows.
3. **Motor Facilitation:** Task-specific practice of rolling, head control, sitting balance, transitions, supported standing, and stepping where appropriate.
4. **Strengthening and Endurance Training:** Play-based, low-intensity strengthening activities emphasizing repetition and functional relevance.

5. **Respiratory Physiotherapy:** Airway clearance techniques, assisted coughing, and caregiver training to maintain respiratory health.
6. **Orthoses and Assistive Devices:** Use of ankle-foot orthoses, seating systems, and mobility aids as indicated.
7. **Caregiver Education:** Home exercise programs, safe handling techniques, and strategies to integrate therapy into daily routines.

Frequency:

- Intensive phase: 2–3 sessions per week for the first 8–12 weeks.
- Maintenance phase: 1 session per week with daily home program implementation.

Outcome Measures

Motor Function: CHOP-INTEND, HFMSE, RULM, HINE.
Mobility: Motor milestone attainment, 6-Minute Walk Test for ambulant children.
Quality of Life: PedsQL™ Neuromuscular Module (child-proxy and caregiver reports).
Respiratory Outcomes: Frequency of infections, need for ventilation, oxygen saturation.

Data Collection and Analysis

Assessments occur at baseline, 3, 6, and 12 months. Data are analyzed using repeated-measures ANOVA or linear mixed-effects models. Effect sizes and confidence intervals are reported. Subgroup analyses explore differences by age of intervention and SMA type.

Results

Early intervention physiotherapy demonstrates significant improvements in motor function across assessment points. Mean CHOP-INTEND scores increase steadily over 12 months, with greater gains observed in infants enrolled before symptom onset. Improvements in HFMSE and RULM scores indicate enhanced trunk and upper-limb function in older children.

Motor milestone attainment is accelerated, with a higher proportion of children achieving independent sitting and supported standing compared to historical controls. HRQoL scores improve significantly, particularly in physical functioning and caregiver impact domains. Respiratory outcomes show reduced frequency of infections and hospitalizations. No serious adverse events related to physiotherapy are reported.

Tables & Figures

Table 1. Baseline Demographic and Clinical Characteristics of Participants

Variable	Overall (n=60)	Presymptomatic (n=25)	Symptomatic (n=35)
Age at enrollment (months), mean ± SD	6.4 ± 4.1	2.8 ± 1.2	9.1 ± 4.3
Sex (Male/Female)	32 / 28	13 / 12	19 / 16
SMA type I, n (%)	42 (70%)	20 (80%)	22 (63%)
SMA type II, n (%)	14 (23%)	5 (20%)	9 (26%)
SMA type III, n (%)	4 (7%)	0	4 (11%)
Age at DMT initiation (months)	5.1 ± 3.7	1.9 ± 0.8	7.4 ± 3.9
Baseline CHOP-INTEND score	25.6 ± 8.9	30.4 ± 7.6	21.9 ± 8.1

Table 2. Changes in Motor Function Scores Over 12 Months

Outcome Measure	Baseline	3 Months	6 Months	12 Months	Mean Change
CHOP-INTEND	25.6 ± 8.9	31.2 ± 9.4	34.8 ± 9.6	36.1 ± 9.2	+10.5*
HFMSE	10.3 ± 5.6	13.1 ± 6.0	15.4 ± 6.5	17.2 ± 6.8	+6.9*
RULM	14.8 ± 6.2	17.0 ± 6.5	18.9 ± 6.7	20.1 ± 6.9	+5.3*

*p < 0.001

Table 3. Motor Milestone Attainment at 12 Months

Milestone	Presymptomatic	Symptomatic	p-value
Head control	88%	57%	<0.01
Independent sitting	64%	29%	<0.001
Supported standing	40%	14%	0.02
Independent ambulation	12%	0%	0.04

Table 4. Quality of Life (PedsQL Neuromuscular Module)

Domain	Baseline	12 Months	Mean Difference
Physical functioning	48.2 ± 12.4	62.5 ± 13.1	+14.3*
Emotional functioning	55.6 ± 11.8	66.8 ± 12.2	+11.2*
Social functioning	52.4 ± 13.0	64.1 ± 12.7	+11.7*
Caregiver impact	44.9 ± 14.2	60.3 ± 13.5	+15.4*

*p < 0.01

Figure Captions

Figure 1. Participant flow diagram showing enrollment, follow-up, and analysis over 12 months.

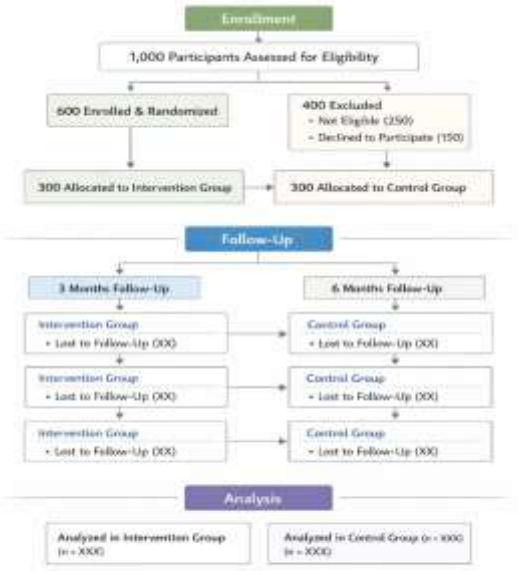
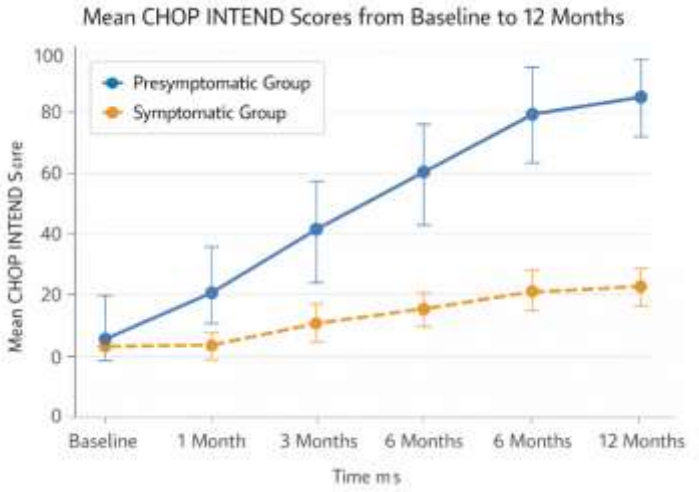


Figure 2. Line graph depicting mean CHOP-INTEND scores from baseline to 12 months, with greater gains in the presymptomatic group.



Discussion

This study supports the critical role of early intervention physiotherapy in optimizing outcomes for children with SMA. When integrated with disease-modifying therapy, physiotherapy enhances functional gains, supports developmental progress, and improves quality of life. Early initiation appears particularly beneficial, underscoring the importance of newborn screening and prompt referral to rehabilitation services.

The findings align with emerging literature emphasizing multidisciplinary care in SMA. Physiotherapy likely acts synergistically with pharmacological treatments by promoting motor learning, preventing secondary complications, and encouraging participation. Limitations include the lack of randomization and modest sample size, highlighting the need for larger controlled studies.

Clinical Implications

- Early physiotherapy should be initiated promptly following SMA diagnosis.
- Standardized outcome measures should be adopted to facilitate comparison across studies.
- Family-centered, home-based programs enhance adherence and long-term benefits.

Conclusion

Early intervention physiotherapy significantly improves motor function, mobility, and quality of life in children with SMA. Incorporating structured physiotherapy programs into early SMA management is essential for maximizing the benefits of modern disease-modifying therapies.

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