Original Article

JHSMR Journal of Health Science and Medical Research

Association Between Gross Motor Functions and Upper Limb Functions in Indian Children with Spinal Muscular Atrophy

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Received 30 June 2025 ● Revised 4 September 2025 ● Accepted 24 September 2025 ● Published online 20 April 2026

Abstract:

Objective: A progressive neuromuscular disease called Spinal Muscular Atrophy (SMA) is characterized by anterior horn cell degeneration, which leads to proximal muscle weakening and compromised motor function. Although upper limb and gross motor skills are frequently impacted, little is known about how these functions interact in the paediatric population of India. The objective of this study was to investigate the relationship between gross motor functions and upper limb functions in Indian children with Spinal Muscular Atrophy.

Material and Methods: Indian children with SMA Types II and III, ages 2 to 18, participated in the cross-sectional study. The Hammersmith Functional Motor Scale Expanded (HFMSE) was used to measure gross motor function, and the Revised Upper Limb Module (RULM) kit was used to evaluate upper limb function. Pearson's correlation test was used to assess the relationship between gross motor and upper limb scores.

Results: A total of 22 participants (mean age: 8.84±5.31; 77% male) were included. A significant positive correlation was found between HFMSE and RULM scores (r=0.579 and p-value=0.005), indicating that higher gross motor functions are associated with higher upper limb functions. The strength of association varied with SMA type and ambulatory status.

Conclusion: In Indian children with SMA, there is a strong association between gross motor functions and upper limb functions. This suggests that both domains should be taken into account in assessment and rehabilitation plans to improve functional status and quality of life in children with SMA. A limitation that can be considered in future studies is to gather data from different regions of India.

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J Health Sci Med Res 2026;44(5):e20261354
doi: 10.31584/jhsmr.20261354
www.jhsmr.org

Keywords: HFMSE, RULM, motor functions, SMA

Introduction

Spinal Muscular Atrophy (SMA) is aneuromuscular disorder with a potentially fatal prognosis[1]. It is characterized by the degeneration of lower motor neurons, resulting in progressive and symmetrical muscle weakness, typically progressing from proximal to distal muscles, including those involved in respiration[1,2]. Clinically, SMA is classified according to the highest motor milestone achieved. SMA Type I, the most severe form and often presenting in early infancy, is characterized by the inability to sit or walk independently. Individuals with Type II may achieve sitting and, in some cases, standing, but do not attain independent walking. SMA Type III presents later in childhood or adulthood, with affected individuals usually retaining the ability to walk without assistance[3,4].

However, the phenotype of survival of motor neuron 1 (SMN1) associated SMA can vary considerably across subtypes. To enhance the quality of future clinical trials, it is essential to use assessment scales that are specifically designed to measure functional abilities and strength-related tasks relevant to SMA. Such tailored scales improve the accuracy and reliability of outcome measurements[5–7]. These assessments should consider factors such as patient age, SMA type, and neurological status, and be conducted routinely preferably every six months by a physiotherapist or physician following a standardized protocol[8].

The Hammersmith Functional Motor Scale Expanded (HFMSE) is one of the most widely used tools for assessing gross motor function in individuals over two years of age who can sit and walk[9]. According to Jessica et al., the HFMSE is particularly suitable for evaluating patients with SMA Types II and III who demonstrate higher levels of motor function[10]. Its strong content validity and clinical relevance for both patients and caregivers make it a robust measure for clinical trials[5]. Another frequently used scale is the Revised Upper Limb Module (RULM), which assesses upper limb strength and fine motor skills in non-sitters, sitters, and walkers aged over 30 months.

Despite the availability of three disease-modifying therapies Nusinersen (Spinraza), Onasemnogeneabeparvovec (Zolgensma), and Risdiplam (Evrysdi) that have improved survival and functional outcomes in SMA, significant challenges remain for patients and their families[11].

Until now, there have been only a few studies that investigated the relationship between upper limb function and gross motor function in SMA patients[12]. Furthermore, there is a considerable gap in the literature regarding studies that directly investigate the relationship between gross motor function and upper limb function in Indian children with SMA. Therefore, this study aimed to examine the correlation between HFMSE and RULM scores among SMA Types II and III, and to compare this relationship between patients receiving disease-modifying therapy and those not receiving such treatment.

Material and Methods

Participants

A total of 22 patients receiving care at the Department of Rehabilitation and Physiotherapy at Sir Ganga Ram Hospital, New Delhi (India), were enlisted for this study. Among these patients, 5 were females and 17 were males, with ages ranging between 2 and 18 years. All individuals had a genetically confirmed diagnosis of SMA, with a homozygous deletion of exons 7 and 8 in the SMN1gene. They were either diagnosed with SMA Type 2 (n=17) or Type 3 (n=5). Out of the 22 patients, 13 were receiving medications such as Nusinersen, Onasemnogeneabeparvovec, or Risdiplam, while the

remaining 9 patients were not receiving any drug treatment. All patients on medications in the study received at least one dose of drug therapy before undergoing the assessment of RULM and HFMSE. None of the patients had unstable medical conditions that would prevent their participation.

Eligibility criteria

Inclusion criteria:

1. Diagnosed case of SMA (Type 2 and Type 3).
2. All patients aged between 2 years and 18 years.
3. All patients are either receiving disease modifying drugs or not receiving drugs.
4. Patients on room air without respiratory support.
5. The caregiver or patient must consent to participate on behalf of their child.

Exclusion criteria

1. Tracheostomy or use of daytime ventilation
2. Acute infections.
3. Acute trauma or fractured cases.

Recruitment

Participants were recruited from Sir Ganga Ram Hospital, Delhi, India.

Patients with a confirmed diagnosis of either SMA Type 2 or 3, referred by a geneticist or neurologist.

Sample size

The Pearson correlation coefficient was 0.579 with a sample size of 22. The calculated G power was 0.839, indicating adequate ability to detect meaningful associations. The sample size for the present study was calculated based on a previously published article, which reported 26 patients[12].

Ethical consideration

This study was approved by the Ethical Committee Review Board of Sir Ganga Ram Hospital on 7th March 2022 (Ref no EC/01/22/1995). The assent form and consent form were signed by the patients and by the caregivers.

Procedure

Two assessment tools, the HFMSE and the RULM, were utilized for evaluation. All assessments were conducted by an occupational therapist in the same order, and both tests were conducted on the same day for each individual.

Outcome measure

HFMSE assesses the gross motor function ordinal scale, which includes 33 items. Each item is scored ordinally from 0–2, with a maximum score of 2 when the item is performed fully without substitutions, 1 with substitutions, and 0 when the item is not performed. The minimum and maximum scores are 0 and 66[5,13]. This scale has demonstrated significant associations in the context of measurement of function, strength, and genotype. Also, according to various clinical trials, HFMSE is a valid, time-efficient outcome measure for SMA Types 1 and 2[5,14].

RULM

RULM assesses upper limb functions; there were a total of 20 items, out of which 18 items were scored with a 3-point system (0-Task not performed, 1-Task performed with modifications, 2-task performed fully) and 2 items were scored with a 2-point system (0-Unable to perform the task, 1-Able to perform the task)[15]. It has demonstrated high inter-rater and intra-rater reliability, and strong construct validity when correlated with other functional scales[16].

Data analysis

The data analysis was performed using the Statistical Package for the Social Sciences (SPSS) version 23.0. Pearson's correlation test was used to analyse the relationship between the various components of the Hammersmith Functional Motor Scale and the Revised UpperLimb Module for spinal muscular atrophy.

Results

Participants

Twenty-two children with SMA Type II and Type III, with 17 children with Type II and 5 children with Type III, were included in the analysis. The mean age of the participants included in the study was 8.84±5.31 years. Out of the 22 participants, 17 were males and 5 were females. Eight children in the study were on Nusinersen, 3 Risdiplam, and 1 Onasemnogeneabeparvovec, while 10 were not on any drugs. Four out of the 20 children were ambulatory, while others were non-ambulatory. The inclusion criteria were children diagnosed with SMA Type 2 and Type 3, age above 2 years, and stable on room air. The exclusion criteria were children suffering from acute infections and children on any respiratory support. The minimum age of onset for the participants included in the study was 6 months. Seventeen children were wheelchair bound. The demographic data for the participants included in the study are given in Table 1. The baseline data for the various components of the Hammersmith Functional Motor Scale and the Revised Upper Limb Module are given in Table 2.

Table 1 Demographic characteristics of the participants (n=22) included in the study

Variable	Mean	S.D.
Age (years)	8.84	5.31
Height (cm)	115.34	23.12
Weight (kg)	24.18	13.55
HFMSE score	23.50	18.08
RULM score	17.36	8.66

S.D.=stanadard daviation, HFMSE=hammersmith functional motor scale, RULM=revised upper limb module

There was a moderate correlation between the Hammersmith Functional Motor Scale scores and the Revised Upper Limb Module, with r=0.579 & p-value=0.005. The scores of the Hammersmith Functional Motor Scale and the Revised Upper Limb Module were significantly correlated with SMA type and ambulatory status (Table 3; Figure 1). The Hammersmith Functional Motor Scale score was significantly correlated with weights, container/lids, and the self-movement components of the RULM scale. Also, the Revised Upper Limb Module score was significantly correlated with transition/crawling, stepping/standing, transition/kneeling, squat/jump, and stairs components of the HFMSE scale (Table 3). There was a moderate correlation between the HFMSE and RULM scores (r=0.584 and p-value=0.046) in children undergoing drug treatment (Nusinersen, Risdiplam, and Onasemnogeneabeparvovec). But there was no significant correlation found in children not undergoing any disease-modifying drug treatment (r=0.569 and p-value=0.086).

Table 2 Baseline data for the various components of the various components of the Hammersmith functional motor scale and the revised upper limb module

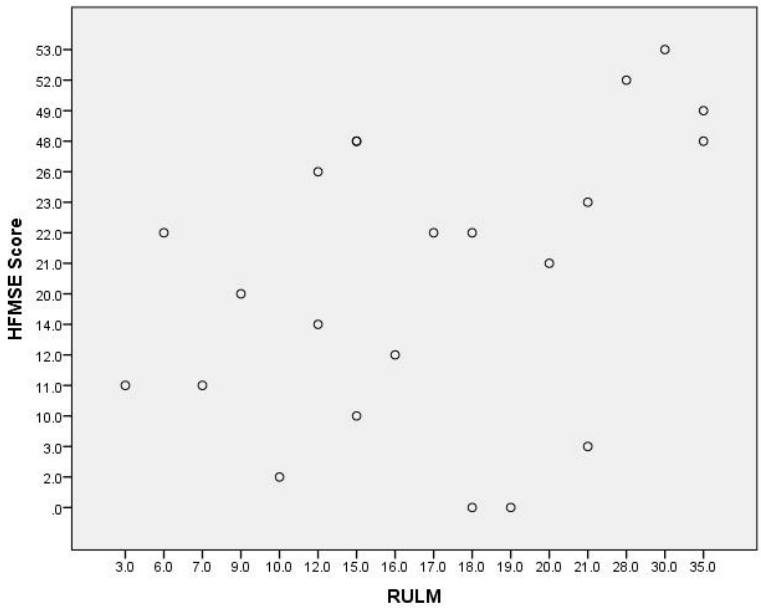
Variable	Mean	S.D.
HFMSE		
Sitting	5.77	2.86
Rolling	7.13	3.77
Transitions/crawling	5.90	6.35
Stepping/standing	1.63	2.73
Transitions/kneeling		
2.31		
3.40		
Squat/jump	0.18	0.58
Stairs	0.54	0.91
RULM		
Paper/pencil	2.31	1.21
Table	1.95	0.21
Coins	5.13	1.32
Plastic cups	1.04	0.65
Push light	0.36	0.58
Weights	4.68	4.33
Container/lids	0.13	0.46
Self-movement	1.72	1.35

S.D.=stanadard daviation, HFMSE=hammersmith functional motor scale, RULM=revised upper limb module

Table 3 Correlational matrix between components of HFMSE and RULM

Variable	HFMSE	Sitting	Rolling	Transitions/ Crawling	Stepping/ Standing	Transitions/ Kneeling	Squat/ Jump	Stairs	RULM	Paper/ Pencil	Table	Coins	Plastic cups	Push light	Weights	Container/ Lids	Self-movt.
HFMSE	–	0.785** (0.000)	.787** (0.000)	0.970** (0.000)	.907** (.000)	0.912** (0.000)	0.519** (.013)	0.907** (.000)	0.579** (0.005)	0.393 (0.071)	0.043 (0.849)	0.246 (0.269)	0.357 (0.103)	0.376 (0.085)	0.683** (0.000)	0.425* (0.049)	0.436* (0.042)
Sitting	–	–	0.912** (0.000)	0.641** (0.001)	.488* (0.021)	0.556** (0.007)	.252 (.258)	0.488* (0.021)	0.298 (0.179)	0.091 (0.688)	−0.174 (0.439)	0.210 (0.347)	0.133 (0.554)	0.52 (0.818)	0.393 (0.070)	0.238 (0.286)	0.217 (0.332)
Rolling	–	–	–	** 0.668 (0.001)	0.476* (0.025)	0.505* (0.016)	0.246 (0.270)	0.476 (0.025)	.255 (.252)	0.053 (0.816)	−0.110 (0.625)	0.140 (0.536)	0.075 (0.741)	0.063 (0.780)	0.355 (0.105)	0.232 (0.299)	0.185 (0.409)
Transitions/ Crawling	–	–	–	–	0.946** (0.000)	0.907** (0.000)	0.514* (0.014)	0.946** (0.000)	0.631** (0.002)	0.481* (.024)	0.102 (0.651)	0.234 (0.294)	0.449* (0.036)	0.435* (0.043)	.725** (.000)	0.469* (0.028)	0.474* (0.026)
Stepping/ Standing	–	–	–	–	–	0.955** (0.000)	0.516* (0.014)	1.000** (0.000)	0.649** (.001)	0.526* (0.012)	0.134 (0.553)	0.252 (0.258)	0.436* (0.042)	0.507* (0.016)	0.721** (0.000)	0.487* (0.021)	0.513* (0.015)
Transitions/ Kneeling	–	–	–	–	–	–	0.588** (0.004)	0.955** (0.000)	0.596** (0.003)	0.414 (0.056)	0.152 (0.499)	0.234 (0.295)	0.358 (0.102)	0.469* (0.028)	0.689** (0.000)	0.421 (0.051)	0.465* (0.029)
Squat/Jump	–	–	–	–	–	–	–	.516* (0.014)	0.435* (0.043)	0.450* (0.036)	0.069 (0.760)	0.212 (0.344)	0.225 (0.313)	0.354 (0.106)	0.546** (0.009)	−0.094 (0.676)	0.185 (0.410)
Stairs	–	–	–	–	–	–	–	–	0.649** (0.001)	0.526* (0.012)	0.134 (0.553)	0.252 (0.258)	0.436* (0.042)	0.507* (0.016)	0.721** (0.000)	0.487* (0.021)	0.513* (0.015)
RULM	–	–	–	–	–	–	–	–	–	0.861** (0.000)	0.216 (0.335)	0.716** (0.000)	0.839** (0.000)	0.767** (0.000)	0.958** (0.000)	0.622** (0.002)	0.879** (0.000)
Paper/Pencil	–	–	–	–	–	–	–	–	–	–	0.243 (0.275)	0.627** (0.002)	0.764** (0.000)	0.708** (0.000)	0.0782** (0.000)	0.424* (0.049)	0.638** (0.001)
Table	–	–	–	–	–	–	–	–	–	–	–	0.023 (0.919)	0.358 (0.102)	0.140 (0.535)	0.138 (0.540)	0.065 (0.773)	0.285 (0.198)
Coins	–	–	–	–	–	–	–	–	–	–	–	–	0.600** (.003)	0.429* (0.046)	0.540** (0.009)	0.200 (0.372)	0.769** (0.000)
Plastic cups	–	–	–	–	–	–	–	–	–	–	–	–	–	0.456* (0.033)	0.745** (0.000)	0.447* (0.037)	0.824** (0.000)
Push light	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.766** (0.000)	0.510* (0.015)	0.557** (0.007)
Weights	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.657** (0.001)	0.765** (0.000)
Container/ Lids	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	0.514* (0.014)
Self-movement	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–	–

*Correlation is significant at the 0.01 level, **Correlation is significant at the 0.05 level
HFMSE=hammersmith functional motor scale, RULM=revised upper limb module



HFMSE=hammersmith functional motor scale, RULM=revised upper limb module

Figure 1 Correlation between the Hammersmith Functional Motor Scale and Revised Upper Limb Module

Discussion

The findings of the present study demonstrated a significant positive correlation between HFMSE and RULM scores in children with SMA. While the HFMSE assesses overall functional ability, the RULM focuses on upper limb strength and fine motor function. This positive association suggests that better gross motor function is accompanied by better upper limb function in this population.

The mean age of the 22 participants was 8.84 ± 5.31 years. The earliest age of onset observed was 6 months in a participant with SMA Type II, whereas the latest was 10 years in a participant with SMA Type III. A significant positive correlation was found between age of onset and SMA type (r=0.491, p-value=0.020), consistent with findings from a large-scale study involving 569 SMA patients[17]. Previous literature indicates that SMA Type II typically presents before 18 months of age, while SMA Type III presents after 18 months[18–20]. This is in agreement with our findings, where the age of onset for Type II ranged from 6 to 18 months and for Type III from 18 months to 10 years.

Of the 22 participants, four were ambulatory—all of whom had SMA Type III—while the remainder were non-ambulatory. This aligns with earlier reports showing that children with SMA Type II are generally unable to walk independently, whereas those with SMA Type III often retain ambulatory ability[21–24]. Twelve participants were receiving disease-modifying therapies [Nusinersen (n=8), Risdiplam (n=3), Onasemnogeneabeparvovec (n=1)], while 10 were not on any pharmacological treatment. A male predominance was noted (male=17, female=5), with more males affected in both SMA Type II (male=14, female=3) and SMA Type III (male=3, female 2). This is consistent with previous studies reporting a higher prevalence of SMA among males[25,26]. The percentage of wheelchair use was higher in SMA Type II (88%) compared to Type III, indicating greater functional impairment in Type II.

Analysis of HFMSE scores revealed that Type II patients had lower scores than those with Type III, reflecting the more severe functional limitations in Type II SMA[27]. Many patients encountered difficulties in tasks requiring greater mobility and complex movements—such as crawling, standing/stepping, climbing stairs, squatting/jumping, and kneeling transitions—likely due to severe contractures, particularly in the knees and hips. Furthermore, the proportion of the maximum possible RULM score achieved (37/44) was slightly higher than that of the HFMSE score (53/66), suggesting relatively better preservation of upper limb function compared to gross motor ability. This observation is in line with Corratti's findings, which reported an average 12-month decline of 0.54 points in HFMSE versus only 0.13 points in RULM[28].

The strong positive correlation between HFMSE and RULM scores observed in our study further supports the utility of these scales in SMA assessment. The HFMSE is a validated and clinically relevant tool designed specifically for SMA[5], providing insights into gross motor capacity[27,30], while the RULM has been shown to be reliable for evaluating upper limb function[14]. Together, these scales provide a comprehensive overview of motor abilities, enabling clinicians to design targeted rehabilitation programs that address both gross and fine motor deficits.

A recent Phase 3b clinical trial known as the SMART study evaluated the efficacy and safety of Onasemnogeneabeparvovec in children with SMA weighing between 8.5 and 21 kilograms. The study found that 61% of participants achieved clinically meaningful improvements in HFMSE scores by week 52, while 41% showed similar gains in RULM, indicating enhanced upper limb function. Importantly, motor function was either maintained or improved across all weight groups, suggesting the therapy's effectiveness even in heavier children. The safety profile was consistent with previous findings, with the most common adverse events being elevated liver enzymes and thrombocytopenia. These results complement real-world evidence and support the broader application of Onasemnogeneabeparvovec in SMA management[31].

In another observational study conducted in South Korea, researchers tracked non-ambulatory SMA patients over a four-year period to assess the long-term impact of Nusinersen treatment. Using a combination of motor function scales—including HFMSE, RULM, CHOP-INTEND, and CHOP-INTEND—the study revealed a consistent positive trajectory in motor function across all tools. Notably, HFMSE scores improved significantly in adolescents, while RULM reflected meaningful gains in upper limb mobility[32].

Clinical implications: Combining HFMSE and RULM in the routine follow-up can enhance the sensitivity of clinical assessments, help tailor physiotherapy and occupational therapy goals, and provide more complete endpoints for clinical trials and registries. Given the heterogeneity in disease course and treatment response, clinicians and researchers should interpret single-scale results in the context of complementary measures (e.g., respiratory function, patient-reported outcomes, and biomarkers) and consider scale limitations such as ceiling/floor effects and age dependency.

However, this study has several limitations. The sample size was small and drawn from a single region in India, limiting the generalizability of findings. Future studies should include larger, more geographically diverse cohorts. Additionally, the sample size of participants receiving disease-modifying therapies was too small to draw robust conclusions when comparing drug-treated and untreated groups. All pharmacological treatments were grouped together under the category of "disease-modifying drugs" without distinguishing between the specific agents or mechanisms of action. This may have masked potential differences in how each treatment influences motor and upper limb function, thereby reducing the granularity of treatment comparisons. Our study was not powered to

detect treatment effects between treated and untreated subgroups; nevertheless, inclusion of treated patients reflects the contemporary clinical practice and underlines the importance of combining gross and upper-limb measures to capture multidimensional treatment effects.

Future research should focus on conducting large, multicenter longitudinal studies in India to capture a more representative SMA population and assess the progression of motor and upper limb function over time. Treatment-specific analyses are also needed to compare the longitudinal effects of individual disease-modifying therapies—such as Nusinersen, Onasemnogeneabeparvovec, and Risdiplam—rather than grouping them together, thereby clarifying agent-specific benefits and optimal initiation timing. Additionally, future studies should incorporate multidimensional assessment frameworks that combine HFMSE and RULM with respiratory measures, patient-reported outcomes, and objective digital tools, enabling a more comprehensive evaluation of functional status and treatment response in children with SMA.

Conclusion

There is a strong association between gross motor and upper limb functions in Indian children with SMA Types II and III.

Author contribution

All authors contributed substantially to the research and preparation of this manuscript. SS conceptualized the study, led data collection, and was primarily responsible for drafting both the initial and final versions of the manuscript. DS contributed to the study design, participated in manuscript drafting, and provided critical supervision and intellectual revision throughout the research process. RDP was responsible for resource acquisition, supported the data collection phase, and provided supervision. RB conducted the data analysis, contributed to the interpretation of findings, and was involved in writing, editing, and preparing the final manuscript. SC provided supervision and strategic guidance across all stages of the study, as well as critical review and revision of the manuscript. All authors have read and approved the final version of the manuscript and agree to be accountable for the integrity and accuracy of the work.

Conflict of interest

None

Funding source

None

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