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Case Report

Multidisciplinary physical rehabilitation program of individuals with spinal muscular atrophy in an inclusive school setting

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Abstract. Spinal muscular atrophy (SMA) is a neuromuscular ailment that leads to the deprivation of motor neurons in the spinal cord, producing denervation and muscle weakness. This case report explains how a patient with type 2 SMA used a therapeutic exercise rehabilitation program in a school environment. Motor functions were assessed by Gross Motor Function Measure-88 (GMFM-88), Manual Muscle Testing (MMT), and Hammersmith Functional Motor Scale (HFMS), which is validated and reliable. This study employed a repeated pre-test post-test measures design. During a year of treatment sessions, the child underwent twice weekly 45-minute physical therapy sessions for 48 weeks. The research was carried out between March 2022 and February 2023. The purpose of the intervention, which comprised a variety of therapeutic workouts, was to enhance physical function and gross motor abilities in an age-appropriate manner. The intervention utilized in this study led to improvements in GMFM-88, HFMS, and MMT total scores. The results of this case study showed that a child with type 2 SMA aged nine had successfully improved their gross motor skills and muscle strength.

Keywords: Spinal muscular atrophy, physical therapy, physical rehabilitation, inclusive education

1. Introduction

Spinal muscular atrophy (SMA) is a neuromuscular condition with an autosomal recessive pattern of inheritance that causes degradation of motor neurons in the spinal cord, resulting in denervation, muscle weakness, and paralysis [1–3]. The "Survival Motor Neuron 1" (SMN1) gene's absence is the causation [4, 5]. One incidence of this illness is seen every 1000 live births [6]. One in 40–60 of the world's population are heterozygotes or carriers [7, 8].

According to the age of onset and the highest level of motor function attained, patients with diverse clinical characteristics can be divided into four phenotypes following the complexity of classical SMA [9, 10]. Type 1 is the most severe type, which appears in the first few months of life. Generalized hypotonia is the earliest symptom. Nearly 90% of children with

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type 1 SMA pass away before the age of two as a result of respiratory issues. In type 2, children can already sit but are confined to a wheelchair permanently and have respiratory issues, contractures, and scoliosis [11]. Type 3 SMA is a milder variant also known as Kugelberg Welander disease. It appears after one and a half years of age. These individuals live, on average, the same length of time as the overall population [12]. SMA type 4 develops after the age of 20. Throughout many decades, type 4 disease symptoms progressively worsen [12, 13].

Type 2 SMA commonly manifests between six and 18 months of age, accounting for 20% of case scenarios. Sitting is typically mastered by nine months, though this developmental milestone may be postponed [14]. Children with type 2 SMA can never walk by themselves but may with the use of a brace or a standing frame. However, some children may be capable of standing. Typically, reflexes are lacking [15]. In type 2, the muscles of the face and eyes are unaffected, but swallowing difficulties, scoliosis, and respiratory insufficiencies are common [16]. Most SMA type 2 patients live to be at least 25 years old, and many live much longer as a result of more intensive supportive therapy [17].

Children with SMA 1 and SMA 2 continue to have a high incidence of scoliosis, with a 60–90% initial presentation rate [10, 18]. Scoliosis more than 20 degrees must be examined on individuals with SMA 1 and SMA 2 every six months until skeletal maturity and once a year after that. Spinal orthoses are frequently used to maintain the hypotonic trunk and cure scoliosis greater than 20° [19]. Individuals with SMA frequently experience hip instability [20]. Reduced range of motion, extended static stance, and agonist-antagonist muscular imbalance all contribute to contractures in SMA patients [20, 21]. Contractures can cause pain and impair function both practically and symptomatically [21, 22].

Multidisciplinary rehabilitation strategies are a crucial element of SMA care [11]. Effective management can change the course of the disease's natural history and lessen its impact [23]. A vital element of rehabilitation health care is assisting people in living as well as they possibly can. A multidisciplinary approach helps to improve physical function, quality of life, and lifespan of those with SMA [24]. In this case, the research team attempted to provide multidisciplinary rehabilitation care.

Inclusive education is a global reform initiative meant to include students with different capabilities in mainstream formal schools [25]. Inclusive education is successfully implemented at both classroom and school levels [26]. Subjects are studying at inclusive school settings and receiving inclusive education as well. Although there is no well-established treatment protocol for SMA management, the research team explored the current evidence-based treatment. The purpose of this case study was to provide an overview of the therapeutic exercise rehabilitation program used for a child with type 2 SMA in an inclusive learning environment.

2. Methods

2.1. Case presentation

A nine-year-old girl with type 2 SMA was brought to an inclusive school by her mother who indicated that she was born at full term, without any issues, and she cried immediately after birth. Her birth weight was reported by her mother to be 3000 grams. The child's main complaints were the inability to sit, stand, or walk freely, as well as weakness in both bilateral upper and lower limbs, with the lower limbs being more evident. Her delayed gross motor skills caused challenges with daily tasks and social interaction. At the age of 2–0 months, she mastered neck control; at the age of 2-3 years, she mastered rolling under supervision; and when she was four years old, she learned to sit with assistance. Before beginning schooling, she did not receive or undergo any physical therapy interventions.

2.2. Ethical approval

The subject for this case study was recruited from an inclusive school. Approval was obtained from the selected school and from the parent. The child and her mother agreed to participate in this observational case report study for 12 months.

2.3. Investigations

Genetic testing that detects deletion of the SMN1 gene and muscular denervation on an electromyography report can diagnose SMA [27]. For this child, it was detected using multiplex ligation-dependent probe amplification (MLPA), which identified positive deletions of exons 7 and 8 of the SMN1 gene and confirmed the diagnosis of SMA, most likely SMA type 2 based on the patient's clinical manifestation. The amount of serum creatine kinase was 111 units

per liter, which was within the biological reference range. The condition was most likely at the AHC level, which causes symmetrical muscular atrophy and weakening.

2.4. Physical interventions

Following examination, the patient was encouraged to visit for routine physical therapy treatment. Short-term and long-term objectives were planned using the problems list as a guide. For the first year, the child had twice-weekly, 45-minute sessions of physiotherapy in addition to twice-weekly, 30-minute sessions of hippotherapy. Physiotherapy will continue to be provided individually, one to three times per week (up to five times per week), in sessions lasting at least 30 minutes until puberty and thereafter at least twice per week until adulthood [28].

2.5. Treatment protocol

During the first four months, therapeutic sessions took place on the mat to attain the anticipated short-term aim. Interventions included stretching exercises, passive range of motion (PROM) exercises to assure the motion range of all joint surfaces, supine to sit facilitation, prone on the elbow, prone on hand, sitting dissociation, and bridging exercises for core strength training [28].

After four months, the same exercises were advanced on the physioball. Treatment also included training for maintaining balance and sustaining seated balance while using a balance board. Hippotherapy with forward reaching and sideward reaching practice was conducted on a weekly schedule with the intent of improving functional activities. To maintain improvement, the same exercises were continued, along with a few additional ones. Therapy sessions commonly started with assisted movement of joint range exercises for joint proprioception to support bilateral weight bearing and weight shifting while sitting. The main goals of therapy in children who are able to sit on their own are preventing contractures and scoliosis, as well as maintaining, restoring, or promoting mobility and functional activity [18].

After eight months, active range of motion (AROM) and strength exercises using manual resistance for the bilateral upper and lower limbs, as well as diaphragmatic breathing exercises, were included in therapy sessions. Independent sitting position practice was started, and finally the subject was able to sustain a sitting position for five minutes without assistance.

Rehabilitation programs can help to maintain and enhance function, strength, joint ROM, stamina, balance, daily living activities, social events, and involvement in school. Exercise programs for children who can sit should include hydrotherapy, eccentric and concentric exercise, aerobic training, and basic conditioning workouts both with and without resistance. Moreover, chest physiotherapy is a crucial component of the examination and care of SMA [18]. Current management entails physical rehabilitation programs twice a week, with each session lasting 45 minutes and including exercises on a balancing board while seated, along with exercises to strengthen upper arms, chest physical therapy, and hippotherapy.

3. Discussion

The purpose of the study was to show the development of a patient with type 2 SMA over one year after receiving a physical therapy rehabilitation intervention. The prime aim of the intervention was to achieve gross motor skills and improve strength in both the upper and lower extremities. After the intervention, improvement was seen in her Gross Motor Function Measure-88 (GMFM-88) score, which went from 29.24% pre-intervention to 37.70% post-intervention. Improvement was seen in A, B, C, and D dimensions. In dimension E, there was no improvement or significant change in pre- and post-intervention scores. In dimension A, improvement was seen in transitional movement, especially while rolling. She was able to lift her head 45° and flex both hip and knee joints through the full range. In dimension B, differences were observed post-intervention, because she was able to sit on the mat and maintain an arm-free position for three seconds. In dimension C, her pre-intervention score was recorded at 28.57%, while the post-intervention score was recorded at 35.71%. In dimension D, the lowest difference was recorded as 0% to 7.69% at pre- and post-intervention, respectively (Table 1).

In a study by Stark et al., patients with type 2 and type 3 SMA experienced significant improvements in the Hammersmith Functional Mobility Scale (HFMS) and GMFM after 12 months of receiving a combination of physical therapy rehabilitation and vibration-assisted sensory motor rehabilitation [29]. In this case, the child's HFMS score improved from

Table 1
Pre- and Post-Intervention scores of Gross Motor Function Measure-88 Scale

Dimension	A (Lying & rolling)	B (Sitting)	C (Crawling & kneeling)	D (Standing)	E (Walking, running, jumping)	Total Score
Pre-intervention	60.78%	56.86%	28.57%	0%	0%	29.24%
Post-intervention	78.43%	66.66%	35.71%	7.69%	0%	37.70%

Table 2
Pre-Intervention and Post-Intervention grades of Manual Muscle Testing

Prime Mover	Pre-intervention	Post-intervention
Elbow Flexor (C5)	Grade 2+	Grade 3+
Wrist Extensor (C6)	Grade 2−	Grade 2+
Elbow Extensor (C7)	Grade 3−	Grade 4
Finger Flexor (C8)	Grade 2+	Grade 4
Finger Abductor (T1)	Grade 2−	Grade 3
Hip Flexor (L2)	Grade 2	Grade 3
Knee Extensor (L3)	Grade 3	Grade 4−
Ankle Dorsiflexor (L4)	Grade 2+	Grade 3
Long Toe Extensor (L5)	Grade 2+	Grade 3
Ankle Plantar Flexor (S1)	Grade 3−	Grade 3+

10/66 to 26/66. The most improvement was seen in the following components: lifting the head from prone, long sitting, supine to side lying, four-point kneeling, and lifting the head from supine.

After the physical therapy rehabilitation program, improvement was also seen in muscle power of both upper and lower extremities using Manual Muscle Testing (MMT) scores. After intervention, massive improvement was observed in the upper extremities, likely the elbow extensor and finger flexor group of muscles. Also there were major recorded changes after physical therapy intervention in the lower extremities, especially the hip flexor and knee extensor (Table 2).

In a study by Lewelt et al., progressive strength training activities were noted to be pain-free, to enhance both motor function and strength, to be practical, safe, and simple to carry out, and well tolerated in individuals with SMA [30]. Gross motor function measurements, statistical muscle testing, tests for pulmonary function, and life quality assessments have been deemed acceptable measurements for clinical trials in young patients with SMA [31]. A previous study showed a substantial improvement with measurements of the GMFM-88, HFMS, and MMT after 1.5 years of physical therapeutic rehabilitation [13].

There is currently no structured rehabilitation strategy for children with SMA available in the literature, which challenged the research team caring for this child. As the condition is rare, the teachers and support staff at the child's school did not have proper knowledge about it. The research team provided continuous support, especially in classroom ergonomics, and encouraged supporting staff and caregivers to give extra care during transfers. Occasionally, the researchers had to confront psychological issues and tried to motivate her to participate in school and the rehabilitation program. The case shows that a multidisciplinary physical rehabilitation program is urgently needed for managing individuals with SMA. In these cases, it is important to have an early diagnosis and to start early rehabilitation to achieve better outcomes.

The research team faced challenges in providing rehabilitation services to the patient with SMA. While delivering care, additional focus needed to be paid to hypermobile joints, ligament laxity, and poor unsteady neck control. There had to be extra caution and concern while providing stretching, weight-bearing, and dynamic balance practice. It was safe for the therapy services to take place on the mat, but it was risky to work on a physioball, on a balance board, or in hydrotherapy. In similar cases, patients should be given proper counseling and motivation about their physical conditions, and encouragement should be given for therapy services. It is also necessary to engage them in academic school activities.

4. Conclusion

Significant positive changes were observed after assessing GMFM-88, HFMS, and MMT scores after one year of completing a goal-oriented adaptive phys-

ical rehabilitation program. The therapeutic exercise rehabilitation program was secure and safe for this child with type 2 SMA. However, extra care and precaution should be taken while providing interventions. It is essential to conduct future research to establish more uniform guidelines for the rehabilitation of children with SMA. Early multidisciplinary intervention should be implemented, individualized, and adjusted for patients with SMA in accordance with their age and condition severity to attain better outcomes.

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Conflict of interest

The authors have no conflicts of interest to report.

Ethical considerations

The patient's mother has given consent for publication.

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