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Role Of The Physical Therapy Intervention In Type 1 Spinal Muscle Atrophy: A Case Study

Samah Al-Harbi, Pediatric PT, Msc, Physical Rehabilitation, KFSHRC

Abstract:
Background: *Spinal Muscle Atrophy Type 1 (SMA1) is a hereditary neuromuscular disorder that affects the motor neuron, leading to sever muscles weakness and limited motor function. Zolgensma therapy has emerged as potential treatment option for SMA1, offering hope for improved outcomes and quality of life for affected infants. However, the effectiveness of the therapy in promoting physical development in SMA1 remains unclear[1]. This case study aim to explore the role of physiotherapy in improving the developmental milestones of an infant with SMA1 following gene therapy. Additionally, will utilize the Chop Intend assessment tool to assess and track the changes in infant's motor function, strength, and over all physical development.*
Case discerption: *7 weeks new borne girl referred from neurology department to physical rehabilitation department, with history of SMA1.*
Management and outcomes: *The baby was seen twice/week, assessed by CHOP-INTEND assessment tool and followed by active ROM exercises, stretching exercises and Neuro-developmental therapy for the last two years. Outcomes was measures by CHOP-INTEND assessment, which improved from 45 to 64.*
Result: *This study highlights the significant benefits of physiotherapy in improving motor milestones in patients with SMA1.*
Keywords: *SMA type 1; NDT; Chop Intend Assessment*

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I. Introduction:

Spinal Muscular Atrophy (SMA) is a genetic disorder that affects spinal motor neurons and is the second most common fatal autosomal recessive disorder after cystic fibrosis[1]. It encompasses a diverse group of disorders marked by phenotypic variability and various genetic mutations [1]. The most prevlent forms of SMA is caused by mutations in the survival motor neuron (SMN 1) gene located on chromosome 5q13 [2]. It is estimated that SMA occurs in 4-10 per 100,000 newborns, making it a relatively rare but highly lethal condition [3 4].

The classification of SMA is based on four main clinical phenotypes (I-V), depending on the age of the onset and the child's maximal functional abilities achieved [5]. Children with SMA1 experience sever weakness and show symptoms within the first six months of life. They are unable to achieve unsupported sitting and may struggle with swallowing and breathing [6]. Unfortunately, many children with SMA 1 do not live past 24 months or require lifelong respiratory support by the age of two [6].

Children with SMA II represents an intermediate and less severe form of the disease. Children with this type are able to achieve sitting and standing abilities, but they never learn to walk without the aid of assistive devices. SMA III is milder and typically manifests after 18 months of age. These children can achieve walking ability independently, and their lifespan does not significantly differ from that of the general population [6]. SMA IV is characterized by its onset after 20 years of age, and symptoms progress slowly over several decades [6].

While there is currently no cure for SMA, significant progress has been made in understanding its molecular genetics and pathophysiology. This has led to the development of several promising therapeutic approaches and preclinical models [1]. In December 2016, three drugs have been approved for the treatment of SMA (Nusinersen, Onasemnogene Abeparvovec-xioi, and Risdiplam). This medication Onasemnogene Abeparvovec-xioi has been shown the most effective therapy.

However, there has been a recent interest in identifying functional measures and appropriate tools, such as the children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND) assessment tool to monitor disease progression. This test assigs a total score ranging from 0- 64, based on the evaluation of various motor skills and movements. The CHOP INTEND allows physiotherapists to gauge different aspects of motor function, including head control, kicking, hand grasp, and even rolling and sitting, providing a comprehensive understanding of an infant's neuromuscular abilities [12]. These SMA 1 infants often struggle to acquire significant milestones as head control, rolling over or sitting position, and they rarely achieve crawling or standing [12].

The aim of the case study is to utilize the Chop Intend as an assessment tool in order to capture developmental milestones changes in the infant with SMA type 1 after receiving the Onasemnogene Abeparvovec-xioi gene therapy. Also to measure the impact of the gene therapy on the developmental milestones of the infants with SMA 1. By tracking and documenting changes in these milestones, the study aims to provide valuable insight in to efficacy of the gene therapy in improving motor function and overall quality of life for infants with SMA 1.

II. Case Report

History

This patient was seen by neurologist at King Faisal Specialist Hospital, Riyadh at age of 6 weeks. Mother reported that the baby was born after an uneventful full-term pregnancy and normal delivery with a birth weight of 2.7 kg, and no perinatal complications happened. However, the positive family history with a prior affected child with spinal muscular atrophy, raises family concerns and prompts genetic testing, which confirmed the homozygous deletion of exon 7 and 8 of SMN1 gene as well as detection of two copies of exon 7 and 8 of the SMN2 gene. This genetic profile is consistent with SMA 1.

During the time, patient is healthy, alert, active and playful but presents with chocking episodes. To ensure safety, a nasogastric tube is inserted. Physical examination revealed that the patient has good upper limbs antigravity movements with good palmar grasp. She was controlling her head in supported siting position. She was able to kick both lower limbs horizontally with eliminated gravity. Due to her age, she was not appropriately yet for rolling. In the prone position, she could not lift the head off the bed. No tightness or contractures were notable.

At the age 6 weeks, patient referred to the pediatric physical therapist with the diagnosis of SMA type 1. The parent reported that the patient's upper and lower extremities movements are weaker compared to her peers. The main parent's complaint was the baby unable to control her head properly, unable to roll and has weakness in both upper and lower limbs however obviously in the lower limbs. Patient did not visit or attempt following physical therapy program during that period.

Additionally, Multiplex Ligation-dependent Probe Amplification (MLPA) Testing for SMA was performed. The MLPA result showed a positive deletion of exons 7 and 8 of the SMN1 gene in homozygous state and detected of two copies of axon 7 and 8 in the SMN2 gene.

Chop intend assessment scale, conducted on 8th of August 2022:

- Scored 45. This highlights the need for early intervention and importance of the physical therapy aimed at maximizing motor function and preventing further complications associated with SMA 1.

Findings:

- Physiotherapy program started after the patient received Zolgensma on 24 August 2021 immediately.
- The patient presented with mild head lag on pull to sit, as well as abnormalities in leg and arm movements.
- Legs were in abducted and flexed, and the arms will move primarily with the elbows on the surface and could not bring it to midline as it shown in figure 1.
- The Chop Intend assessment score was measured and scored 45.
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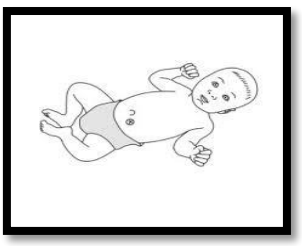

Figure 1: Frog position

Plan of care:

- PT twice/week and HEP was provided
- Active ROM, stretching exercises for upper & lower extremities.
- Milestones stimulation program were also introduced to encourage head control by doing pull to sit exercises, rolling to both side, and encourage weight bearing on both upper extremity from the prone position. These exercises were to be performed daily as the patient tolerated. As it shown in Figure 2, 3, and 4.

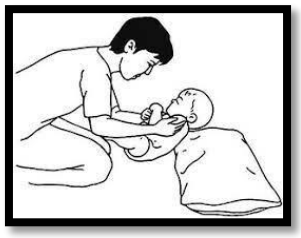

Figure 2: Pull to sit exercise

Figure 3: Rolling to both sides

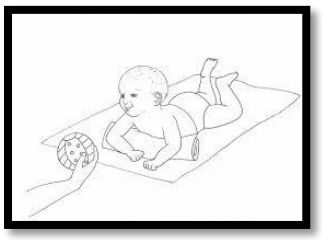

Figure 4: Tummy time

Outcomes:

- *At age of 4 months,* patient's parent reported minimal improvements in head control and upper extremity movements.
- The Chop Intend assessment was measured and improved to 52

New plan of care:

- Instructed the parents to adhere with the home exercises program.
- Maintain active ROM and stretching exercises, as well as the pull to sit and rolling to both sides, and encouraging tummy time exercises.
- *At age of 7 months,* the parent noticed increased lower extremity strength and the patient started sitting with maximum support as it shown in Figure 5.
- The Chop Intend assessment score increased to 54.

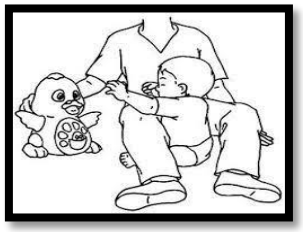

Figure 5: Sitting with maximum support

New plan of care:

- The physiotherapist recommended continuing with the physiotherapy program as above.
- Introduced exercises such as sitting exercises by using proper chair, encourage pivoting exercises, trunk exercises with a Swiss ball as it shown below in Figure 6, 7, and 8.
- Standing exercises using a standing frame for 30 minutes daily.
- The joint proprioception exercises were also implemented for bilateral lower limbs in Tall-kneeling position.

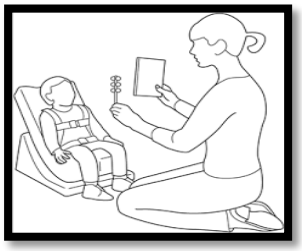

Figure 6: using proper chair

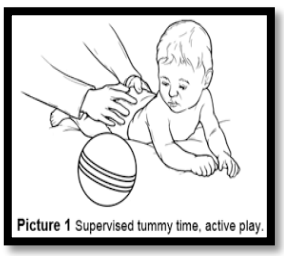

Figure 7: Pivoting

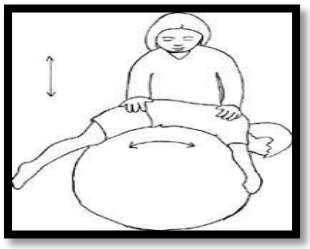

Figure 8: Trunk exercises

- *At age of 15 months,* the parent noticed significant improvements, including independent sitting, walking with a baby walker, and standing at the edge of the bed with two hands support.
- Parents concern raised regarding the development of bilateral pronated feet as it shown in Figure 9.

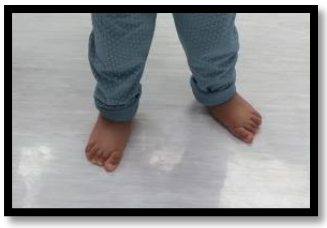

Figure 9: Pronated feet

- The Chop Intend assessment score improved to 58.

New plan of care:

- Physiotherapist advised continuing with the program, incorporating crawling exercises as it illustrated in Figure 10, proper shoe usage as it illustrated in Figure 11 , cruising exercises, and transition exercises via moving from sofa to table and vice versa.
-

Figure 10: Crawling exercise

Figure 11: Medical shoe

- *At age of 19 months,* parents expressed their satisfaction with their child's progress, noting that the child had begun crawling and cruising independently and was able to walk with two hands support.
-
- The parents was concerned that the child was still unable to attain the sitting position from a lying posture alone, necessitating parent's assistance.
- Chop Intend assessment score was utilized, and scored 60. This score indicated the child's level of motor function and overall progress.

New plan of care:

- The physiotherapy exercises previously prescribed were continued.
- Several exercises involving joint proprioception in the bilateral lower limbs were incorporated into the treatment plan. These exercises included Tall-knelling exercises , pull to sit movement from the floor as it shown in Figure 13, and standing against a wall with appropriate footwear.
- The child was encouraged to perform static standing balance exercises to improve her ability to maintain balance in a stationary position.

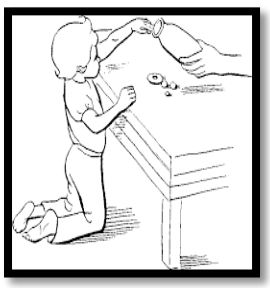

Figure 12: Tall-knelling exercises

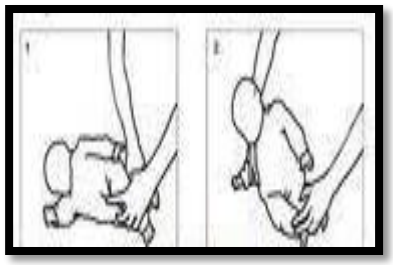

Figure 13: Sitting from sideway

Figure 14: Walking with support

At age of 20 months, the parents expressed their satisfaction with the child's progress, as the child had begun taking steps independently with close supervision.

- The parent also expressed concerns about the child's walking pattern, particularly the presence of bilateral pronated feet.
- The Chop Intend assessment score was the same previous visit 60.

New plan of care:

- The physiotherapist introduced a few exercises specifically aimed to improve the child's walking ability. These exercises included rising up from floor and climbing stairs, which would help strengthen the muscles involved in walking.
- The parent was advised to ensure that the child wore appropriate shoes to improve her feet posture. It was explained to parent that proper footwear could contribute to better alignment and reduce the pronation of the feet.
- The parent was advised to seek a referral to an orthotics specialist. They would be able to provide custom-made Ankle Foot Orthosis support as showed in figure 16. It inserted in the shoes in order to correct the pronation and provide additional stability for walking.
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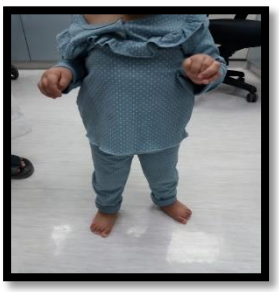

Figure 15: Patient is walking independently

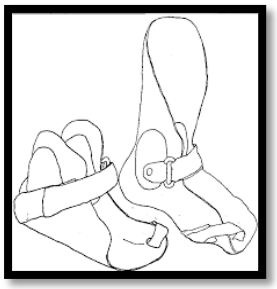

Figure 16: AFO

At age of 2 years, parent reported that the child started to walk independently at home and the child had also managed to climb stairs with support of a rail.

- These achievements not only indicated the child's physical progress but also demonstrated her increased strength and mobility.
- The Chop Intend assessment score developed to 64.

III. Results

Preliminary results showed evidence of positive changes in SMA1 infant milestones and muscle strength following Gene therapy in conjunction with physical rehabilitation intervention.

The results demonstrated that The Chop Intend assessment score was improved from 45 to 64 through two years of follow up as it shown at the below table.

Table 1: The Post Intervention Score of Chop Intend assessment:

Items	Chop Intend Assessment
Spontaneous movement (Upper Limb)	20%
Spontaneous movement (Lower Limb)	16.6%
Hand grip	20%
Head in midline with visual stimulation	20%
Hip adductors	20%
Rolling elicited from legs	15.8%
Rolling elicited from arms	17.5%
Shoulder and elbow flexion and horizontal abduction	18.3%
Shoulder flexion & elbow flexion	16.6%
Knee extension	18.3%
Hip flexion and foot dorsiflexion	16.6%
Head Control	17.5%
Elbow flexion score with Item 14	20%
Neck flexion score with 13	18.3%
Head/ Neck extension (Landau)	13.3%
Spinal Incurvation (Gaiant)	10%

IV. Discussion

This case study explored the impact of the physical therapy in improving motor milestones in the patient with SMA 1 over a period of two years after receiving Zolgensma therapy. The main focus of the study was to achieve gross motor development, gain proper posture, and improve muscles strength in both upper and lower extremities. To capture possible changes in the achievement of motor developmental milestones in infancy, Chop Intend scale was utilized as a clinical tool to assess motor function in weak patients with neuromuscular disorders including infants with SMA 1. This scale has been widely used in multiple natural history studies of SMA type 1, as well as in multiple SMA clinical trials in order to identify the variability of the phenotype and assess the rapid progression of the disease.

Previous studies have been shown that infants with SMA1 are unable to achieve independent sitting or other motor milestones such as crawling and standing [16]. However, the finding of the current study was inconsistent with these previous results as the patient showed significant progression after the intervention. The patient's motor skills and muscles strength showed improvement, as reflected in the Chop Intend Scale. The total post-intervention score increased by 17.4%, with improvement seen in all items of the scale, including upper and lower muscles strength, head control, and rolling abilities.

Based on this finding of the current study, the study strongly encourages regular physical therapy sessions for all SMA1 patients, at least 5 times a week depending on the patient's general health status. Previous studies have also shown that infants with SMA 1 who receive regular physiotherapy are able to perform movements and achieve motor milestones. Therefore, it is crucial to prioritize physiotherapy as part of the overall treatment plan for SMA 1 patients [17].

V. Conclusion

This study highlights he significant benefits of physiotherapy in improving motor milestones in patients with SMA1. By incorporating regular physiotherapy sessions into the treatment plan, patient can experience improvements in motor skills, muscle strength, and overall quality of life. The importance of physiotherapy cannot be overstated, and it should be considered an integral part of care for SMA1 patients.

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