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Case Report Open Access

Use of Therapeutic Exercises in Type II Spinal Muscle Atrophy: A Case Report

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Abstract

Spinal muscular atrophy (SMA) is a neuromuscular disorder characterized by degeneration of alpha motor neurons. This case report describes use of therapeutic exercise rehabilitation program with type II SMA. Motor skills were examined using the Goss Motor Functional Measures (GMFM)-88, Hammersmith Functional Motor Scale (HFMS) and Manual Muscle Testing (MMT) which are reliable. The child received Physical Therapy rehabilitation program on regular basis for 40-min sessions for 18 months. The intervention included various therapeutic exercises and it was designed to improve gross motor skills and age appropriate functional mobility. Improvement was seen in total score of total score of GMFM-88, HFMS and MMT by intervention given in this study. The outcomes of this case report demonstrated that, the successful improvement of gross motor functions and strength in muscles in 3-year-old child with type II SMA.

Keywords: SMA; Therapeutic exercises; GMFM-88; HFMS; MMT

Abbreviations SMA: Spinal Muscular Atrophy; GMFM-88: Goss Motor Functional Measures-88; HFMS: Hammersmith Functional Motor Scale; MMT: Manual Muscle Testing; SMN: Spinal Motor Neuron, AHC: Anterior Horn Cell

Introduction

Spinal Muscular Atrophy (SMA) is the commonest serious autosomal recessive disorder after cystic fibrosis. In 1890s this disease was first described by Werdnig and by Hoffmann. Incidence of SMA is 1 in 6,000 to 1 in 10,000 live births [1]. Spinal muscle atrophy is defined as a genetic disorder of the motor neurons in the anterior horns of the spinal cord resulting in degeneration of the spinal cord alpha motor neurons [2]. The cause of SMA is deletion or mutation of survival motor neuron 1 (SMN1) which encode the SMN protein [3]. On the basis of age onset and achievement of motor functions SMA is classified into four types [1].

Type I SMA is the most sever type and it is also known as Werdnig Hoffmann disease. Onset of disease occurs within six months after birth. These patients never achieve independent sitting. The lifespan of infants with SMA type I is less than 24 months without ventilator support [3].

Type II SMA is an intermediate as well as less severe form. It is also known as Dubowitz disease. The onset of disease occurs between 6 months and 18 months of age [2]. Patients are able to sit independently and some of them are able to maintain standing position, but they are unable to walk independently [1]. They live up to adulthood age [3].

Type III SMA is milder form and is also known as Kugelberg Welander disease. Type III SMA occurs after 18 months of age. The average lifespan of individuals do not differ from the general population [1].

The onset of SMA type IV is after 20 years of age. The symptoms of type IV disease gradually progress over numerous decades [1].

SMA is identified by genetic testing in which deletions of the SMN-1 gene and EMG report shows denervation of muscles [2]. There are various outcome measures for SMA such as Motor Function Measure-32 (MFM), Gross Motor Functional Measures-88 (GMFM-88), Quantitative muscle testing, Hammersmith Functional Motor Scale (HFMS), pulmonary function test, pedsQL etc. [4-6] (Tables 1, 2 and 3).

Motor function in children with type II SMA assessed by using GMFM-88 scale [7]. The HFMS was modified for determination of functional ability in children with SMA and MMT is tool used to assess the strength of muscle [2,8].

Case Report

History

This patient was 1 and half year old female brought to Pediatric Physiotherapy Department by mother with the medical diagnosis of Type II Spinal Muscular Atrophy. Her mother reported not as such any significant birth history or complications (full term normal vaginal delivery, immediate cry was present after birth, birth weight was 3000gm). Chief complaints were unable to coming to sitting from supine lying, unable to sit independently and weakness in bilateral upper and lower extremities but it was more prominently in lower extremities. Her gross motor development was delayed. She achieved neck control at the age of 6 months, rolling achieved at the age of 10 months. She did not received or attempt any physical therapy treatment session before this.

Investigations

Multiplex ligation-dependent probe amplification (MLPA) Testing for Spinal Muscular Atrophy (SMA) showed positive deletion of exon 7

and 8 of SMN1 gene with the diagnosis of Spinal Muscular Atrophy likely SMA type II on the clinical presentation. EMG report showed that, chronic motor axon degeneration affecting proximal and distal upper limb and lower limb muscles. Site of lesion is likely to be at AHC level.

Intervention

After evaluation referred to orthotic and prosthetic department for orthosis and to genetic counselor for genetic counseling they are advised to come for regular Physiotherapy treatment. On the basis of problems list, short term and long term goals were planned. Initially Child was on Physiotherapy treatment session for 5 days per week for 45 minutes almost up-to 2 and half years of her age was given.

Treatment protocol

First 6 months, session began with exercises on mat. Intervention included, passive range of motion (PROM) exercises to bilateral upper and lower limb, supine to sit facilitation from both sides, prone on elbow, prone on hand, sitting dissociation and combination of quadruped position and abdominal curl ups for core muscle strengthening.

Then same exercises were progressed on Swiss ball. Treatment also included working on maintaining and tolerating sitting balance on wobble board, reach out exercises in sitting and the patient achieved sitting independently at the age of 2.

Same exercises were continued for maintenance and few more exercises were added. Session typically began with active assisted range of motion exercises (AAROM), Joint proprioception exercises to bilateral lower limb by giving NDT techniques like kneeling, half kneeling, pull to stand from floor, standing on one leg, stride standing, standing against ball with and without ankle foot orthosis (AFO). She achieved standing independently at 2.8 years of age.

After that, session would begin with active range of motion (AROM) and strengthening exercises with manual resistance to bilateral lower extremities, diaphragmatic breathing exercises. Initially walking started with the help of walker then it was progressed on treadmill and she was able to maintain standing position without support for 10 min.

The current episode of care involves physical therapy rehabilitation sessions 3 days in each week and the duration of daily session is 40 minutes in which balance board exercises in standing, bilateral lower limb strengthening exercise, sit to stand, treadmill walking is included and also mother is advised to continued same exercises protocol at home daily.

Results

A (Lying & Rolling)	B (Sitting)	Dimension	C (Crawling & Kneeling)	D (Standing)	E (Walking, Running & Jumping)	Total Score
56.80%	0%	Pre-Intervention	0%	0%	0%	11.36%
96.07%	98.33%	Post-Intervention	92.85%	41.02%	22.22%	70.09%

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

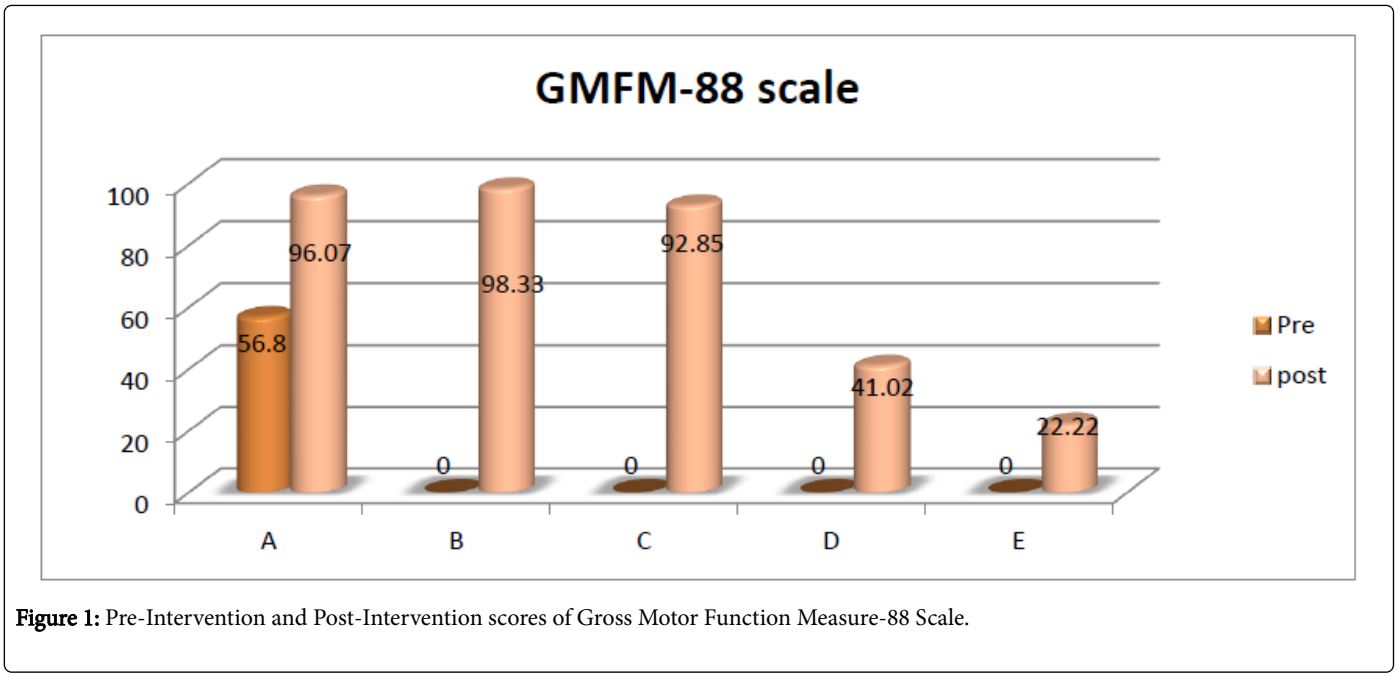

Figure 1: Pre-Intervention and Post-Intervention scores of Gross Motor Function Measure-88 Scale.

Pre-Intervention score	Post-Intervention score
8/66	45/66

Table 2: Pre-Intervention and Post-Intervention scores of Hammersmith Functional Motor Scale.

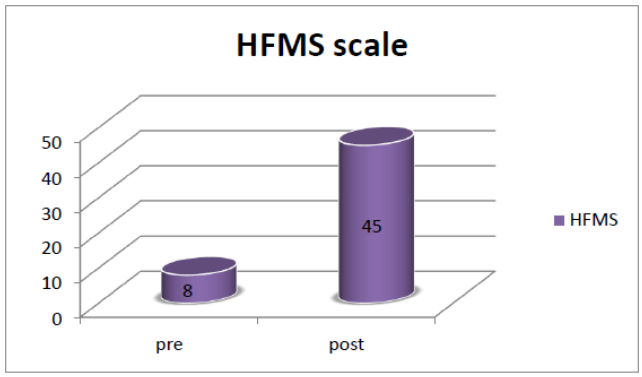

Figure 2: Pre-Intervention and Post-Intervention scores of Hammersmith Functional Motor Scale.

	Pre-Intervention assessment		Post-Intervention assessment	
	Right	Left	Right	Left
Hip				
Flexors	Grade 3	Grade 3	Grade 4	Grade 4
Extensors	Grade 1	Grade 1	Grade 3	Grade 3
Abductors	Grade 1	Grade 1	Grade 3	Grade 3
Adductors	Grade 2-	Grade 2-	Grade 3	Grade 3
Internal rotators	Grade 1	Grade 1	Grade 3	Grade 3
External rotators	Grade 1	Grade 1	Grade 3	Grade 3
Knee				
Flexors	Grade 3-	Grade 3-	Grade 4	Grade 4
Extensors	Grade 3	Grade 3	Grade 4	Grade 4
Ankle				
Plantar flexors	Grade 1	Grade 1	Grade 3	Grade 3
Dorsiflexors	Grade 1	Grade 1	Grade 3	Grade 3
Invertors	Grade 1	Grade 1	Grade 3	Grade 3
Evertors	Grade 1	Grade 1	Grade 3	Grade 3

Table 3: Pre-Intervention and Post-Intervention grades of Manual Muscle Testing.

Comments from the mother

According to the mother, she had been greatly benefited from the physical therapy rehabilitation program. Her strength, balance and walking has improved. She was able to move more independently on floor than before since they started Physical Therapy treatment session.

Discussion

The purpose of study was to see the development in patient with type II SMA over a period of 1 and half years after receiving the Physical Therapy rehabilitation intervention. The prime aim of the study was to achieve gross motor skills and improve the strength in bilateral lower extremities. After the intervention improvement was seen in GMFM-88. Total pre-intervention score was 11.36% and total post-intervention score was 70.09%. Thus, the improvement was seen in all A, B, C, D, E dimensions. In dimension B, improvement was seen in sitting balance she was able to touch the toys behind 45°. In dimention C, she was able to crawl backward down 4 steps on hands and knee, able to attained forward 10 steps kneel walking with free arm. In dimention D, she started maintained static standing balance for 20 sec with arm free. In dimention E, she was able to ascend and descend 4 steps with holding one sided railing.

Christina Stark et al. concluded that, patients with type II and type III by giving combination of Physical Therapy rehabilitation and vibration assisted neuromotor rehabilitation there was significant positive changes occurred after 12 months in Hammersmith Functional Mobility Scale and Gross Motor Function Measure 66 [9] (Figures 1 and 2).

Improvement was seen in Hammersmith Functional Motor Scale (HFMS) and it was improved from 8/66 to 45/66. Most improvement was seen in following components sitting independently, four-point kneeling, crawling, High kneeling to left and right half kneeling, unsupported standing, initiates squats with arm support, ascends and descends 2-4 stairs with railing support (Table 2).

After physical therapy rehabilitation programmed improvement is also seen in muscle power of bilateral lower extremities. Aga Lewelt et al. reported that, progressive resistance training exercises program was pain free and improvement in motor function as well as in strength was seen and also these exercise program was practicable, safe, easy to perform and well tolerated in children with SMA [10].

Sussan T. Iannaccone et al. said that, Gross Motor Function Measures, quantitative muscle testing, pulmonary function tests and quality of life were reliable outcome measures for clinical trials in pediatric patients with spinal muscular atrophy [4].

Conclusion

The therapeutic exercises rehabilitation program was safe in patients with types II spinal muscular atrophy. Significant beneficial changes occurred after measuring the GMFM-88, HFMS and MMT after 18 months after receiving goal-oriented functional physical therapy rehabilitation and home-based exercises.

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Conflicts of Interest

The authors report no conflict of interest. The authors alone are responsible for the content and writing of this paper.

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