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Research Article

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Effects of Individualized Intensive Physical Therapy for a Child with MECP2 Duplication Syndrome: A Case Study

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| Abstract |

PURPOSE: High-intensity physical therapy may help improve some gross motor developmental delays through an increased treatment frequency. This study describes an increase in physical therapy frequency and intensity over an eight-week time frame for a child with a rare genetic neurodevelopmental disorder.

METHODS: A single-subject research was performed. The subject was a nine-year-old boy with MECP2 Duplication Syndrome. The outcome measure consisted of one time before and after the intervention. An intensive physical therapy program was applied to this subject. The treatments included reciprocal crawling, high kneeling, tailor sitting, weight-bearing and shifting training to facilitate ankle balance strategies and training to walk without assistance.

RESULTS: After the eight-week follow-up, the child achieved a gradual improvement in the gross motor function measures, and the mother reported that the child frequently engaged in tailor sitting and independently walking.

CONCLUSION: This case study highlights that intensive physical therapy improved the overall motor function of a child with MECP2 duplication syndrome.

Key Words: Case reports, Genetic diseases, Physical therapy modalities

Ⅰ. Introduction

MECP2 duplication syndrome (MDS) is a rare X-linked neurodevelopmental disorder characterized by a wide variety of symptoms, including early onset low muscle tone, mental retardation, developmental delays, frequent respiratory infections, absent speech, seizures, midface hypoplasia, depressed nasal bridge, large ears, progressive spasticity, and ataxic gait [1]. Among these characteristics, a decline in functional development due to developmental delay or low muscle tone can significantly reduce the child's quality of life and worsen the symptoms due to secondary degenerative musculoskeletal problems. According to previous research, children with cerebral palsy (CP) also face difficulties because of similar issues, and it has been reported that

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the role of physical therapy (PT) is crucial [2].

PT plays a leading role in managing pediatric rehabilitation. It concentrates on the remediation of physiological deficits and on ameliorating functional limitations and participation restrictions. PT uses emerging therapy approaches to improve, retain, and recover biological, psychological, and social health. In addition, it educates caregivers on how to manage children at home with regard to the activities of daily living.

Research on CP patients has shown that an intensive PT program improves the outcomes of PT, regardless of the type of PT [3]. Increasing the frequency of treatments from twice to four times a week has been shown to improve motor abilities, as measured by the gross motor function measurement (GMFM) scores [4]. These results confirm that more intensive PT can expedite motor skill acquisition in impaired children with low muscle tone. On the other hand, there are no studies s of PT for children with MDS. This case study describes the PT management of a child with MDS using intensive PT.

Ⅱ. Methods

1. Patient History

The patient was a nine-year-old boy weighing 28 kg. He could recognize and respond to the faces of people he frequently saw but exhibited avoidance behavior towards unfamiliar individuals. Although he could not speak, he responded to others by observing their behavior through facial expressions and actions. He was not taking any medication at the time. He was the only child of unrelated healthy parents, born at term by a cesarean section after intrauterine growth retardation. He was born with hypotonia, measuring 46 cm in height and 2.2 kg in weight. Motor developmental delay was evident from the age of four months. He achieved head control at six months, trunk control at 11 months, creeping at three years, and crawling with bunny hopping at four years. He could walk with a walker, but could not stand or walk alone. The mother stated that he had many problems in infancy. In particular, he did not grow well, showed generalized muscle weakness, and suffered recurring severe respiratory infections that required hospitalization. On the other hand, he had never presented with seizures.

In December 2015, he was diagnosed with MDS, a syndrome characterized by duplications of the Xq28 region encompassing the methyl-CpG binding protein 2 (MECP2) gene. Since the diagnosis, he has received PT twice a week at rehabilitation hospitals for the delayed development of his gross motor function. The PT sessions usually took 20 to 30 minutes, depending on the hospital. On the other hand, he could not receive PT from 2020 to 2022 due to COVID-19. After the cessation of therapy, he rarely attempted to stand independently. In addition, when placed in an upright position by a caregiver, he showed hesitation in sitting down and demonstrated signs of functional decline.

2. Initial Impression and Movement Observation

He came into the room holding his mother's hand. When he first entered the room, he did not make eye contact and crawled straight to the window with a bunny-hopping motion. The mother reported that the child struggled with reciprocal crawling and interacted poorly. He could not actively weight shift and reciprocally dissociate the lower and upper extremities because crawling with bunny hopping adopts bilateral symmetrical movement of the lower and upper extremities. The mother also reported that he always tried to sit in a W-sitting position (WSP) when playing on the floor. When he encountered the sofa while bunny hopping to the window, he could climb up onto it. The mother said that climbing the sofa is an activity that he enjoys. On the other hand, he showed symmetrical extremity movements while climbing up on the sofa. With two hands on the furniture, he could rise to stand through

to a high kneeling position. He never showed himself standing or walking. His mother said that he could not stand alone for more than a second, and he appeared to be afraid.

During the child's PT examination, his mother said that she was concerned about his gait ability hindering his participation in the daycare center. She expressed her desire for him to be able to gait with his friends. Written consent was obtained on an informed consent form. This case study focuses on the goal related to gait ability, and the study was conducted ethically according to the principles of the Declaration of Helsinki [5].

3. Pre- and Post-Intervention Testing

Pre- and post-intervention testing was conducted by a PT practitioner with five years of experience with the gross motor function measure (GMFM-88) at the Y Children Development Center. The evaluator was unaware of the research content. He contacted the subject's guardian to schedule an assessment date with the child. The assessment was conducted in an evaluation room without any observers present. The tests included two components of the International Classification of Functioning, Disability, and Health (ICF) [6]. The ICF activity component was measured using the GMFM-88, and the participation component was measured from an interview with his mother. The GMFM has 88 items; each was scored on a four-point ordinal scale from “does not initiate (score 0)” to “completes the movement (score 3).” The scores for the five dimensions were measured: (A) lying and rolling; (B) sitting; (C) crawling and kneeling; (D) standing; (E) walking, running, and jumping. The GMFM score is highly reliable, valid, and sensitive to change [7]. The reliability of GMFM-88 was above .99. The validity was indicated to conform to the assumption of unidimensionality [8]. The responses for the guardian interview were provided through simple oral questions rather than a standardized questionnaire.

4. Intervention

The intensive PT program consisted of five sessions per week, two hours per session, for eight weeks. Intensive PT was delivered at the Y Children Development Center. Intensive PT was completed by one therapist trained in intensive PT intervention, with 15 years of experience in children with CP. He attended the intensive therapy program held by the instructor, Kevin S. Huang, in Taiwan from August 15 to August 19, 2016. Implementing intensive therapy of two hours per session, five days a week, for eight weeks may potentially be a strenuous regimen, particularly considering the characteristics of the child. Therefore, the number of sets and rest intervals were adjusted based on the child's condition. Each intervention session combined two components:

(1) The functional training component focused on guiding the child to achieve postural stability and balance through normal development-related functions, such as reciprocal crawling, high kneeling, tailor sitting, standing alone, balance training, and walking training [9].

(2) The caregiver education component consisted of guidance for the parent or assistant on handling and home training within the limit of daily activities at home [10]. The home program was structured with the interventions performed, including reciprocal crawling and transitioning from sitting to standing without support, all conducted in the clinic setting. The mother directly observed the treatment, received verbal instructions, and was guided to perform similarly. She was instructed to follow along similarly.

The child's favorite music and videos were played on a tablet PC during all treatment sessions. Many toys and tools were also used to motivate the child in functional activities, balance training, and walking training.

The functional training exercises were designed to target the development of postural stability and balance through antigravity movements: reciprocal crawling, transfer from sitting on the heels to high kneeling, tailor sitting on the

Table 1. Parameters for functional training exercises

Parameter	Intensity	Volume	Target goal	Frequency/ Duration	Rest	Note
Reciprocal crawling	30 minutes	Repeating a 10m distance 5 times	Improve shoulder and core postural stabilization	Five sessions per week / 8 weeks	1 – 2 minutes after every 10 meters	Train for differentiated movements before crawling
Training for tailor sitting on a stability ball	15 minutes	3 sets, with 5 minutes per set	Contract the elongated hip external rotation muscles	Five sessions per week / 8 weeks	1 - 2 minutes after every 10 meters	Changes the position of the ball continuously
Transfer from sitting on the heels to high kneeling	15 minutes	10 sets of 20 - 40 repetitions	Control of both gluteus maximus muscles	Five sessions per week / 8 weeks	1 - 2 minutes between sets	Guide to preventing forward bending of the spine
Standing balance	30 minutes	3 sets, with 10 minutes per set	Improve balance while standing using the ankle strategies	Five sessions per week / 8 weeks	1 - 2 minutes between sets	Guide to shift the weight towards the heels
Gait training	30 minutes	Repeating a 10m distance 5 - 10 times	Improve independent walking	Five sessions per week / 8 weeks	1 - 2 minutes between sets	Minimizing the assisting force

ball, independent standing, and gait training (Table 1). The goal of reciprocal crawling is to improve shoulder and core postural stabilization, weight-bearing (WB), weight shifting (WS) on the upper extremities (UE) and lower extremities (LE), and strengthening the elbow extensors, hip flexors, and extensors (Fig. 1). For reciprocal crawling, he was able to lift his trunk from the floor while WB and WS on UE and LE. After this training, he took sitting on the heels as a starting position. He transferred his posture from sitting on his heels to high kneeling (Fig. 2). The training goal was to promote postural stability and balance of a high kneeling position through functional skills and improve the control of both gluteus maximus muscles. The goal of training in the tailor sitting posture was to stretch

Fig. 1. Training for differentiated movements before crawling.

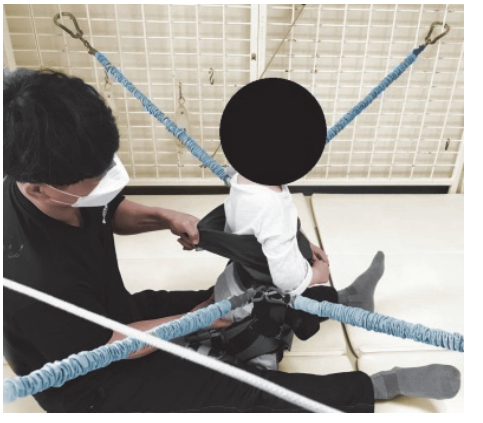

Fig. 2. Transfer from sitting on the heels to high kneeling.

Fig. 3. Training for tailor sitting on a stability ball.

Fig. 4. Training for independent standing on a balance board.

the shortened hip internal rotation muscles and contract the elongated hip external rotation muscles (Fig. 3). To improve the child's standing balance strategy, he was trained for 30 minutes to use the ankle strategy on the movable balance board, using a movable platform capable of anterior-posterior displacements or dorsi-plantar flexion of the ankle joint (Fig. 4). The type of perturbation is an environmental condition that can affect the balancing strategy. The balancing response can be triggered by sensory input from an unpredicted perturbation (feedback activity) [11]. Finally, for independent gait, 30 minutes of gait training was conducted using an elastic cord (elastic cord, 8mm, YAMUZIN, Korea). An elastic cord provided the child with as little assistance as possible.

Table 2. Score for GMFM before and after the intensive PT (IPT)

Variables	Baseline	After IPT[a]
Total score	154	201
lying and rolling	51	51
Sitting	47	59
Crawling and kneeling	33	39
Standing	11	27
Walking, running, and jumping	12	25

[a]Intensive physical therapy

Ⅲ. Results

Table 2 lists the GMFM-88 scores of the child on the five dimensions of the GMFM. A higher score indicates better gross motor function. All dimensions of the GMFM-88 were increased except for dimension A (lying and rolling), which was already 100%. The child could walk approximately 9 m (30 ft) by himself. The quality of his walking pattern showed asymmetry. He had less control over his right leg and took shorter steps.

An interview with his mother showed that her child participated more in activities without WSP, sideward cruising with one-hand support along the furniture, and participated in more programs at the daycare center. In addition, her child appeared to be more active when it came to playing with other children, and he appeared to have developed psychological confidence.

Ⅳ. Discussion

MDS is a rare genetic neurodevelopmental disorder characterized by symptoms, such as hypotonia, delayed development, and spasticity from muscle stiffness aggravated by voluntary movement and can be related to involuntary muscle contractions [12]. The number of PT

programs designed specifically for children with MDS is limited. Hence, effective PT programs for children with MDS are needed.

This case study describes the application of intensive PT to a child with MDS. Although a case study lacks the control of a research study, other studies have documented the importance of using contemporary practice knowledge [13,14]. This case study shows that an intensive PT intervention administered for eight weeks in a child with MDS improved the gross motor function, as measured with the GMFM. This finding corresponds well with the results of intensive PT studies in CP [15,16]. The child showed high levels of adherence to the intensive PT program and demonstrated evidence of progression.

The child developed an ability to walk approximately 9 m (30 ft) by himself. Walking is a complex activity. Muscle weakness can contribute to walking disability. Other factors, such as motor planning, standing balance, and postural control, can affect the walking function. Consistent with the specificity of training, the child with MDS appeared to improve what he practiced (mat exercise, standing balance, postural control, and gait training). In designing the training regimen for children with MDS, aligning the exercises and activities with their specific functional goals and challenges is essential. This approach ensures that the training is tailored to address the unique needs and abilities of each child. For example, if a child's primary goal is improving gait patterns, the training program should incorporate exercises directly targeting the gait mechanics. This methodology, consistent with the principle of specificity of training, emphasizes the importance of tailoring interventions to the specific objectives of the individual, leading to more effective and meaningful progress in their functional abilities [17].

After the intensive PT program, the child's time spent in the WSP decreased significantly, while the time spent in the tailor sitting position increased. The WSP is the flexion of the knee joint and flexion, adduction, and internal rotation of the hip joint. Children who can sit independently using WSP often demonstrate tightness of the hip flexors, adductors, internal rotators, and hamstring muscles. The WSP also contributes to the poor development of the pelvic-femoral muscles. When the child played in the WSP, his lumbar spine and rib cage rotated over his pelvis. His pelvic rotation did not occur over the femur. He used his pelvis for postural stability. If the WSP pattern is used over long periods, it decreases trunk and hip control.

The present case study has important clinical implications. The case study is a method of inquiry through which physical and occupational therapists can inform practice and provide foundation knowledge for clinical trials [18]. The rare neurodevelopmental disorder encompasses many conditions with few patient representatives [19]. In that respect, case study in PT proves even more valuable. This case study demonstrates how current clinical practice can be used with a child with a rare genetic neurodevelopmental disorder. This is believed to be the first study of MDS associated with intensive PT.

The limitations of this case study include the limited generalizability of the study results. There is no control for the maturation threat to internal validity, and an influence other than the independent variable (intensive PT) may have caused the changes in the dependent variables (motor function). Multiple baseline assessments would have allowed a comparison to determine whether the improvement in results was due to natural maturation. The caregiver interview was evaluated based on responses to simple verbal questions rather than a standardized questionnaire. Therefore, it was unreasonable to assess the 'Participation' component of the ICF based on this. More studies will be needed to examine the effects of intensive PT on children with MDS.

Ⅴ. Conclusion

This case study should remind pediatric physical therapists that intensive PT can improve the gross motor function of children with MDS. The clinical decisions for a child with MDS in contemporary practice could be based on the physical therapist's management.

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