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THE RESULTS OF ENVIRONMENTAL ENRICHMENT AND NEURODEVELOPMENTAL TREATMENT IN SHPRINTZEN-GOLDBERG SYNDROME: A CASE REPORT

ORIGINAL ARTICLE

ABSTRACT

Purpose: This case report aimed to investigate the efficacy of the physiotherapy program in a child with Shprintzen-Goldberg syndrome (SGS).

Methods: A 9-month-old case diagnosed with SGS by a medical geneticist was the focus of the study. Congenital pes varus, craniosynostosis and craniofacial deformities, mental, social, emotional and motor retardation, regulation and sensory hyperreactivity symptoms were observed in the case. The physiotherapy program consists of environmental enrichment and Neurodevelopmental Treatment (NDT) approaches twice a week for 12 months and was conducted by a physiotherapist at Karadeniz Ereğli Private Gökkuşağı Special Education and Rehabilitation Center. Gross motor function and disability level were assessed using Gross Motor Function Measurement-88 (GMFM-88) and Gross Motor Function Classification System (GMFCS), respectively. The success of physiotherapy goals was determined by Goal Attainment Scaling (GAS).

Results: At 12 months follow-up, GMFM-88 score increased from 5.52% to 45.47% and GAS total scores increased from -6 to +6 point.

Conclusions: The literature review shows that no previous study assessed the effectiveness of 12 months physiotherapy program in a child with SGS. We consider that physiotherapy including targeted NDT approaches can support motor development in rare cases, such as SGS characterized by severe motor involvement.

Keywords: Case Report, Craniosynostosis, Multiple Anomalies, Physical Therapy and Rehabilitation, Shprintzen-Goldberg Syndrome

SHPRİNTZEN-GOLDBERG SENDROMUNDA ÇEVRESEL ZENGİNLEŞTİRME VE NÖROGELİŞİMSEL TEDAVİ SONUÇLARI: BİR OLGU SUNUMU

ARAŞTIRMA MAKALESİ

ÖZ

Amaç: Bu olgu sunumu, Shprintzen-Goldberg sendromlu (SGS) bir çocukta fizyoterapi programının etkinliğini araştırmayı amaçlamaktadır.

Yöntem: Tıbbi genetik uzmanı tarafından SGS tanısı konmuş 9 aylık bir olgu çalışmaya dahil edildi. Olguda konjenital pes varus, kraniyosinostoz ve kraniyofasiyal deformiteler, mental, sosyal, emosyonel ve motor retardasyon, regülasyon ve duyu hiperreaktivite semptomları gözlendi. Haftada iki gün, 12 ay boyunca çevresel zenginleştirme ve Nörogelişimsel Tedavi (NGT) yaklaşımlarını içeren fizyoterapi programı, Karadeniz Ereğli Özel Gökkuşağı Özel Eğitim ve Rehabilitasyon Merkezi'nde fizyoterapist tarafından uygulandı. Kaba motor fonksiyon ve özürlülük düzeyi sırasıyla Kaba Motor Fonksiyon Ölçümü-88 ve Kaba Motor Fonksiyon Sınıflandırma Sistemi ile değerlendirildi. Fizyoterapi hedeflerinin başarısı, Hedefe Ulaşma Ölçeklendirmesi ile belirlendi.

Sonuçlar: On iki aylık takipte, Kaba Motor Fonksiyon Ölçümü-88 puanı %5,52'den %45,47'ye, Hedefe Ulaşma Ölçeklendirme toplam puanları -6'dan +6 puana yükseldi.

Tartışma: Bu olgu sunumu, literatür taramasına göre SGS'li bir çocukta 12 aylık fizyoterapi programının etkinliği hakkında yapılan ilk çalışmadır. Hedefe yönelik NGT yaklaşımlarını içeren fizyoterapinin ciddi motor etkilenim ile karakterize SGS gibi nadir görülen olgularda, motor gelişimi destekleyebileceğini düşünüyoruz.

Anahtar Kelimeler: Çoklu Anomaliler, Fizik Tedavi ve Rehabilitasyon, Kraniyosinostoz, Olgu Sunumu, Shprintzen-Goldberg Sendromu

INTRODUCTION

Shprintzen-Goldberg syndrome (SGS) is an extremely rare connective tissue disorder involving several body regions, first documented by Shprintzen in 1982 (1). It is characterized by craniosynostosis of coronal, sagittal or lambdoidal sutures, dolichocephaly, typical craniofacial features, neurological findings, mental retardation and skeletal abnormalities such as scoliosis, joint hypermobility or contractures, pectus chest deformity, multiple abdominal wall hernia and infantile hypotonia (2,3).

Although there is no complete cure for SGS, a lifelong multidisciplinary approach is recommended due to developmental delay and the presence of cardiovascular anomalies (4). In addition to other treatment approaches, it was recommended that physiotherapy interventions for deformities should begin as early as possible (2). To our knowledge, there are no studies examining cases of SGS involving developmental delays that show the results of physiotherapy intervention as prognostic in the long term.

The aim of this study is to demonstrate the effectiveness of physiotherapy intervention and to discuss the results of 12 months follow-up in a child with SGS. We consider that this study will contribute to the literature on the long-term (12 months) results of physiotherapy intervention in individuals with SGS.

CASE PRESENTATION

The 9-month-old baby was admitted to the rehabilitation center by his family with the complaint of hypotonia, i.e., the inability to sit or roll. The baby had not previously received physiotherapy intervention. The birth weight of the baby was 3750 g, the gestational age was 38 weeks, and the delivery method was cesarean section. For the Auditory Brainstem Response scan, results were normal limits of hearing function in the left ear, and mild conductive hearing loss in the right. According to the genetic test results, it was concluded that the c.104C>G (p.Pro35Arg) variation detected in the SKI gene was consistent with Shprintzen-Goldberg syndrome and clinical findings. He the second child of the family that had no history of genetic disease. The genetic structure of the family is being investigated. The baby was taking no medication. He has a five-year old sister. He did not need neonatal intensive care after birth.

According to medical reports, he had bilateral flexible congenital varus deformity of the foot, craniosynostosis, craniofacial deformities, mental retardation, pectus chest deformity, high palate,

Table 1. The Goals Steps of The Case

GAS	-2	-1	0	+1	+2
GAS-1	He cannot roll from supine to prone.	He can roll from supine to side-lying position.	He can roll from supine to prone	He rolls over a distance of 3m from supine to prone, and from prone to supine.	He rolls over a distance of 10m from supine to prone, and from prone to supine.
GAS-2	He cannot sit without support.	He can sit with his hands supported from the front.	He can sit without support.	He can move his trunk in the sagittal and frontal planes in the sitting position without support.	He can move his trunk in the vertical plane in the sitting position without support.
GAS-3	He cannot play with toys in the sitting position.	In a sitting position, he reaches for the toy in front of him with his hands but falls.	He can reach for the toy in front of him and play with his hands in a sitting position.	He can reach and play toys of different heights in the sagittal and frontal planes in the sitting position.	He can reach and play with toys with trunk rotation in sitting position.

GAS: Goal Attainment Scaling

GAS-1: Rolling while lying, GAS-2: Sitting without support, GAS-3: Playing with toys while sitting.

umbilical hernia, and undescended testis. The baby's regulation skills were quite weak and mostly lethargic. He had poor midline orientation, body awareness, and avoided tummy time positions. He was unable to bear weight on his legs or sit without support. It was observed that he did not use his extremities for functional skills such as reaching, playing with toys, or rolling. He also had sensory reactivity problems, such as avoiding touching the floor with his feet and hands in supine, prone, and supported sitting positions. He is not comfortable with movement and position transitions. In daily life, he is unable to roll, reach out, sit or stand, i.e., his participation level is limited.

Written informed consent was provided by the child's parents.

Physiotherapy Assessments

(1) Disability level: We used the Gross Motor Function Classification System (GMFCS) to measure the level of disability. The classification levels range from I (independent ambulatory function but some minor troubles of balance, speed or coordination) to V (no independent ambulatory function). The GMFCS is an accepted classification system used to classify motor functions in cerebral palsy and other pediatric problems (5).

(2) Gross Motor Function Measurement (GMFM): We used GMFM-88 for evaluating gross motor function of the case. This tool is suitable for children with ages ranging from 5 months to 16 years. The child's movements and postures are benchmarked with specific identifiers and these are used to determine change in performance over time. The GMFM-88 item scores can be summarized to count raw and percent scores for each of five GMFM category, selected target areas and a total GMFM-88 score (6).

(3) Goal Attainment Scaling (GAS): GAS is a method for individual goal-setting. In a study, 70% of therapists and 60% of parents confirmed that GAS is an appropriate tool to improve rehabilitation quality (7). Goal settings were edited by parents, and the therapist according to the child's areas of interest. The goals were considered SMART, an acronym for specific, measurable, achievable, relevant and timed. The achievement of goals was rated using a scale of five points (-2 to +2), with 0 equivalent to goal achievement. The scores means that '–2' is the initial pre-treatment (baseline), '–1' is progression towards the goal without attainment, '0' is the expected level after treatment, '+1' is a better-than-expected outcome, and '+2' is a much better-than-expected outcome. Three of the goals created by SMART were selected: rolling while lying, sitting without support, and playing with toys

Table 2. Gross Motor Function Measurement-88 And Goal Attainment Scaling Results

GMFM-88	Pretreatment	Posttreatment
A. Lying and Rolling (%)	17.64	100
B. Sitting (%)	10	60
C. Crawling and Kneeling (%)	0	57.17
D: Standing (%)	0	10.25
E: Walking, Running and Jumping (%)	0	0
Total score (%)	5.52	45.47
GAS (point)		
1. Rolling on lying	-2	+2
2. Sitting without support	-2	+2
3.Playing toys on sitting	-2	+2
Total GAS score	-6	+6

GMFM-88: Gross motor function measurement-88, GAS: Goal Attainment Scaling

GMFM-88 data expressed as a percentage (%).

while sitting. These three goals were chosen to create the transfer skill of the child, to develop postural control against gravity, to increase the upper extremity functions in the sitting position and to support the playing skills (Table 1).

Treatment

The physiotherapy program started when the case was 9 months old. The case was evaluated in Kdz. Ereğli Private Gökkuşağı Special Education and Rehabilitation Center between February 2021 and February 2022 and was included in the rehabilitation program during this period. The duration of the treatment was two days a week for 12 months, each session being 60 minutes. The case was assessed and treated by a pediatric physical therapist with at least 10 years of experience (S.A.T). The treatment concept was NDT approach and environmental enrichment therapy (Figure 1, Figure 2). In the NDT concept, techniques such as facilitation of extensor muscles, facilitation of rolling, righting and balance reactions were practiced with the use of appropriate handling techniques and key points. These studies were integrated into the child's rehabilitation and home environment according to the principles of environmental enrichment. For example, one of the goals was sitting without support, and by creating a safe environment, i.e., placing him in a basket, it was possible to develop the righting and balance reaction in sitting. The family were present at the physiotherapy sessions. A home program was given for its implementation in line with the determined goals; the child received no additional treatment.

Also, orthoses were used for correct alignment of the lower extremities. Since the patient was standing with bilateral hip-knee flexion, ring-locked Knee Ankle Foot Orthosis was given to ensure proper lower extremity alignment. He used the orthosis for 5 hours daily. No adverse effects were experienced during the therapy process.

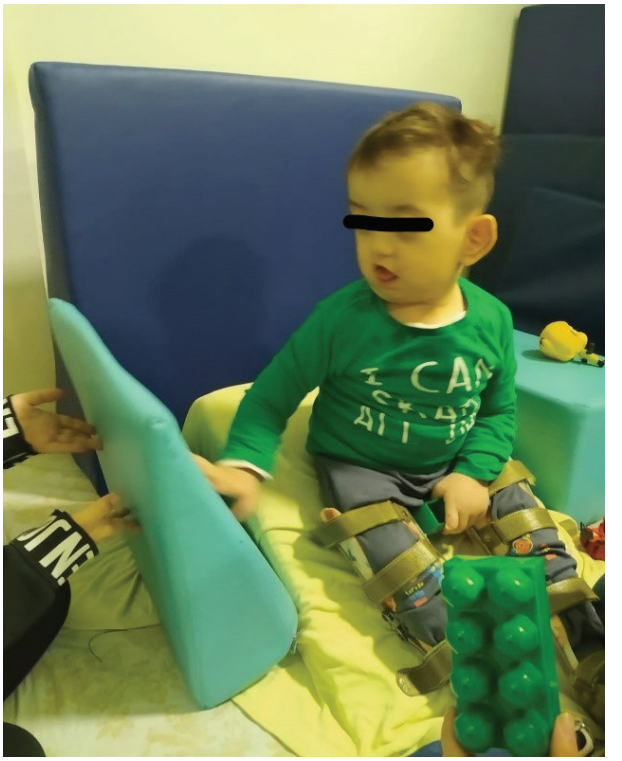

Figure 1. Playing Toys on Sitting Position with Enriched Environmental Perspective

Figure 2. FFunctional Reaching on the Kneeling Position According to the Neurodevelopmental Treatment Approach

Outcome and Follow-Up

At the end of the 12-month follow-up period, our case was 21 months old. During this time, GMFCS levels changed from level V to level II.. The percentage of total GMFM-88 and GAS scores before and after treatment are shown in Table 2. GMFM-88 increased from 5.52% to 45.47% and GAS total scores increased from -6 to +6 point.

DISCUSSION

SGS is a rare genetic disorder with specific features. The features in this case are consistent with previous cases reported in the literature (2,3). A child's overall quality of life and participation can be improved by a multidisciplinary approach that includes input from a physician, cardiologist, pediatrician, otorhinolaryngologist, ophthalmologist, surgeon, speech and language pathologist, physiotherapist, radiologist and a pediatric dentist (4). In addition to its use primarily in cerebral palsy, NDT has also been used as a supportive rehabilitation technique in other conditions (8). NDT is a holistic problem-solving approach based on the teaching of normal movement patterns based on the evaluation and treatment of impairment in function (9). Environmental factors can sometimes be an obstacle for children but can also have a facilitating effect. It is known that development is affected by individual and environmental factors, and recent studies have often emphasized the effects of the environment, whether complicating or facilitating, on the function and disability process of children with physical disabilities (10). In the literature, the definition of environmental enrichment includes socialization, exercise, sensory and cognitive stimulation and environmental modification. This approach emphasizes that the individual's environmental, social, physical and cognitive context should encourage positive stimulation, interactions and activities, and this positive experience will stimulate, support and contribute to the development of structural changes in the brain due (11). Therefore, for our case we selected the NDT approach and enriched environmental therapy. In this case, the NDT approach supported the child's postural alignment, and environmental enrichment provided the child with the opportunity to move more independently. With this combined approach, more movement was achieved while maintaining postural alignment.

Well-defined goals need a goal-oriented scale as a measure of rehabilitation success. There are four main goals of rehabilitation: the target activity, the support needed, quantification of performance and the time for achievement (12). GAS is a personalized assessment scale that measures progress towards defined goals. It is a common evaluation method in the field of physical therapy and rehabilitation, in which setting goals is a fundamental part of treatment planning (13). The key aspect of this case report is the goal-oriented basis of rehabilitation program through the use of SMART principles. The family and the physiotherapist agreed on the methods used and various roles taken in achieving these goals. In this case, to determine the achievement of rehabilitation goals, we used the GAS, which is convenient, quick, patient-specific, goal directed and widely applicable. In this case, the three SMART goals were selected, for each of which better-than-expected outcome scores were found after 12 months of NDT approach and environmental enrichment therapy.

Although the typical and clinical features of SGS have been described in previous case reports (2,3), due to the lack of specific tests to determine the levels of motor function and disability, we used three different functional outcome measures in our evaluation: GMFCS, GAS and GMFM-88. After the NDT approach, according to these measurements with proven validity, the level of change in motor function and disability levels is described as improvement, no change, or worsening. In this case, we used GMFCS to assess the disability level. The patient's GMFCS level progressed from level V before treatment to level II after treatment. This indicates that the NDT approach positively affects the gross motor function level of a patient with SGS, and leads to an improvement in disability level.

Gross motor function was assessed by GMFM-88 (8). GMFM-88 has been mainly used to evaluate children with cerebral palsy in the literature (14,15), but also to evaluate the motor functions of children with other diagnoses, such as Down syndrome (16) and muscular dystrophy (17). Two different studies reported that NDT approach is

useful for lying, rolling, sitting, crawling and kneeling, and standing abilities but there was no significant improvement in walking, running and jumping in children with cerebral palsy (14,15). Similarly, in this case, we determined that after the treatment, there was improvement in all four abilities, except for walking, running, and jumping. The GMFM-88 scores of our case, who is 21 months old is as follows: can turn and lie down, 100%; can sit, 60%; can crawl, 57.17%; can stand, 10%.

In this study, the environmental enrichment and NDT approach allowed the child to develop in social, emotional, motor, and sensory areas. It was observed that the child, who had had no experience of social interaction lethargic and compatible after the 12-month therapy process. We are therefore of the opinion that the development of motor skills also contributes to the child's emotional well-being. The child's poor regulation skills negatively affected his participation in life. Environmental enrichment and NDT approach contributed to the child's participation in therapy, and the experience of acting independently; applying the principle of 'just right challenge', therapy involved movement experience, and games supporting all development areas (social, emotional, motor, cognitive). 'Just right challenge' offers activities that are sufficiently challenging, and this principle results in faster progress in areas targeted for development (18).

We consider that the combination of NDT approach and environmental enrichment therapies were beneficial gross motor skills and increased the level of functional independence for this case with SGS. In addition, it is important to support all the child's developmental areas (social, emotional, cognitive, motor), integrating therapy into all areas of life, and family education.

In conclusion, SGS is a rare connective tissue disease with multiple anomalies, and for a child with SGS, physiotherapy is an important part of a multidisciplinary teamwork aimed at reducing joint deformities, functional disability and increasing the level of motor function. To the best of our knowledge, this case report is the first study on the effectiveness of physiotherapy and rehabilitation program in a child with SGS. The physiotherapy results were encouraging in this case with SGS. However, this study is a case report, making it difficult to generalize the results, which is the limitation. Additional studies and randomized controlled trials are needed to confirm these benefits and the efficacy of a physiotherapy intervention for cases with SGS in long-term follow-up.

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