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Results of Neurodevelopmental Therapy in Galactosemia: A Case Report

Galaktozemide Nörogelişimsel Terapi Sonuçları: Olgu Sunumu

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Öz

Amaç: Erken süt çocukluğu döneminde nörolojik semptomları farkedilen olguda 8 haftalık fizyoterapi-nörogelişimsel tedavi ile ilgili verilerimizi sunmayı amaçladık. **Yöntem:** Galaktozemi tanısı ve tipik genetik yüzü olan 39 haftalık 3400 gr olarak dünyaya gelen kız hasta 11. ayında desteksiz oturamama, ayakta duramama ve yürüyememe şikayetiyle fizyoterapi programına alındı. Hastanın demografik verileri kaydedildi. Tedavi öncesi ve sonrasında motor gelişim düzeyi, kas tonusu ve refleksel gelişimi değerlendirildi. Motor gelişimini değerlendirmek için Klasik motor değerlendirme ve Kaba Motor Fonksiyonel Sınıflandırma Sistemi (KMFSS), kas tonusu değerlendirilmesi için Modifiye Ashwort Skalası (MAS) ve palpasyon, refleksel gelişimi için denge ve düzeltme reaksiyonları kullanıldı. Olguya 8 hafta süreyle haftada 2 gün Nörogelişimsel Terapi (Bobath Terapisi) uygulandı. **Bulgular:** 8 haftalık Nörogelişimsel tedavi sonrası; baş kontrolü, dönme ve alt ekstremite hareketlerinde artışla birlikte motor gelişim düzeyinde gelişme görüldü. Düzeltme ve denge reaksiyonlarında ve reflekslerinde düzelme saptandı. **Sonuç:** Galaktozemi gibi metobolik sendroma bağlı gelişim geriliği olan bebeklerde erken dönem nörogelişimsel terapinin uygulanması motor gelişim açısından oldukça önemlidir.

Anahtar kelimeler: Galaktozemi, Bobath Terapisi, Fizyoterapi

Abstract

Aim: In this study, the results of 8-week physiotherapy-neurodevelopmental treatment were presented in the case whose neurological symptoms were noticed in early infancy. **Method:** A 39-week-old girl with a diagnosis of galactosemia and a typical genetic face of 3400 g, was admitted to the physiotherapy program at the 11th month with complaints of inability to sit, stand and walk without support. Demographic data of the patient were recorded. The level of motor development, muscle tone and reflexes were evaluated before and after the treatment. Classical motor assessment and Gross Motor Function Classification System (GMFCS) were used to evaluate motor development, Modified Ashwort Scale (MAS) was used to evaluate muscle tone and palpation, reflex assessment was used for Correction and equilibrium reactions. Neurodevelopmental Therapy (Bobath Therapy) was applied to the case 2 days a week for 8 weeks. **Results:** After 8 weeks of Neurodevelopmental treatment; Head control, rotation and lower extremity movements increased, as well as improvement in motor development level. Correction and balance reactions and reflexes were also better. **Conclusion:** The application of early neurodevelopmental therapy is very important in terms of motor development in infants with developmental delay due to metabolic syndrome such as galactosemia.

Keywords: Galactosemia, Bobath Therapy, Physiotherapy

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1. Introduction

Galactose-1-phosphate is a disorder of galactose metabolism due to uridyl transferase deficiency. While classical galactosemia manifests itself with clinical findings such as poor sucking, growth retardation, jaundice, and bleeding diathesis in the neonatal period, it results in life-threatening complications such as feeding problems, growth retardation, bleeding, hepatocellular damage in untreated infants. Definitive diagnosis is made by determining the deficiency of galactose 1 phosphate uridyl transferase activity. It is autosomal recessive. While its incidence is between 1/40.000-1/80.000, it is seen at a rate of 1/23.775 in our country (1, 2). There are no studies on the efficacy of physiotherapy in these patients with growth retardation due to its rarity. We aimed to present our data on neurodevelopmental treatment for 8 weeks in a patient whose neurological symptoms were noticed in early infancy.

2. Case Report:

A 39-week-old girl with a diagnosis of galactosemia and a typical genetic face of 3400 g, was admitted to the physical therapy outpatient clinic with complaints of inability to sit, stand and walk without support in the 11th month. It was stated that the case had convulsions in the 8th month and had epileptic seizures.

The pre- and post-treatment evaluation of the case was started with demographic evaluation. Anthropometric measurement (circumference) method was used for head circumference measurement. Classical motor assessment and Gross Motor Function Classification System (GMFCS) were used to determine the motor development level of the case. Palpation and Modified Ashwort Scale (MAS) were used to assess muscle tone. Reflex assessment was performed to evaluate her reflex development.

Neurodevelopmental therapy (Bobath) was applied to the case 2 days a week for 8 weeks. According to the classical motor evaluation stages; In the case with poor head control, flexion head control was positive while extension directional head control was insufficient. Rolling from supine to prone, prone to supine was negative, and the case could only be prone from positioned semi-lying. It was noted that there was no supported sitting, crawling and walking, and that the trunk balance was weak. According to GMFCS 5 Lower extremity movements (Flexion, extension, abduction, adduction) against gravity were insufficient.

Eye tracking was recorded as negative in our case with swallowing weakness. According to the reflex development evaluation; Correction and equilibrium reactions were found to be negative. In the evaluation of muscle tone; It was determined that it was 0 according to the MAS and there was a decrease in tone with palpation. Deep tendon reflexes (Achilles, patella, biceps, brachioradialis, triceps) were decreased. Also, head circumference was determined as 42.5 cm in anthropometric measurement.

At 8 weeks post-treatment evaluation While eye tracking started after treatment, there was no change in swallowing difficulty. According to the classical motor evaluation stages; Head control, leg movements (Flexion, extension, abduction, adduction), turning (from prone to supine, from supine to prone) and supported sitting were positive, while crawling and walking were negative according to GMFCS. According to the reflex development evaluation; An increase in muscle tone (normal) was observed. The deep tendon reflexes Achilles and brachioradialis were bilaterally normal, and correction and balance reactions were developed. Anthropometric measurement; Head circumference was determined 45 cm as well.

3. Discussion

Galactosemia is one of the autosomal recessive disorders of carbohydrate metabolism. Although galactosemia is observed in all three enzymes in galactose metabolism, the most common GALT enzyme deficiency is encountered (1). Ozturk et al. (4) reported the rate of consanguineous marriage as 57.1% in their study. In the light of all these data, autosomal recessive The fact that this inherited metabolic disorder is more common in our country than in developed countries may be associated with consanguineous marriages. Raising public awareness and The necessity of preventing consanguineous marriages is clear. But in our case, the parents are not relatives.

A child with galactosemia is normal at birth. Clinical

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findings such as inability to gain weight, vomiting, diarrhea, and lethargy occur after galactose intake. Afterwards, jaundice, hepatomegaly, abnormal liver function tests, cataracts, and renal dysfunction begin to appear (5,6) In addition, symptoms related to the neurological system appear as developmental delay, progressive psychomotor retardation, convulsions, and disorders of the central or peripheral nervous system (6,7)

When these children reach school age, they have reading difficulties and their neurological problems increase (8). For this reason, we think that early diagnosis and support with physical therapy is important in terms of child's motor development.

Our case had syndromic facial appearance, developmental delay, hypotonus and epileptic seizures. In the physiotherapy evaluation, in the case with motor development retardation, insufficient correction and balance reactions, improvement was observed in the level of motor development, correction and balance reactions and reflexes after NDT Bobath application. Early application of neurodevelopmental therapy is very important in infants with developmental delay due to metabolic syndromes.

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Conflict of Interest

The author declare no potential conflicts of interest with respect to the research, authorship and/or publication of this article.

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