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Case report: physical therapy and rehabilitation process of a patient diagnosed with achondroplasia

Olgu sunumu: akondroplazi tanılı hastanın fizik tedavi ve rehabilitasyon süreci

Sevda CANBAY DURMAZ[1] Ümran GÜVEN[2] Ali CANBAY[3] Davut ÖZBAĞ[4]
Selma SOLGUN DAĞ[5]

[1] Mardin Artuklu University, Faculty of Medicine, Department of Anatomy, Mardin, Türkiye.
[2] Zirve Special Education and Rehabilitation Center, Malatya, Türkiye.
[3] Malatya Battalgazi State Hospital, Malatya, Türkiye.
[4] Adıyaman University, Faculty of Medicine, Department of Anatomy, Adıyaman, Türkiye.
[5] İnönü University, Institute of Health Sciences, Department of Anatomy, PhD Program, Malatya, Türkiye.

ABSTRACT

Achondroplasia is an autosomal dominant disorder. But 80% of cases are sporadic. Its incidence is between 1/25000 and 1.5/10000. Significant shortening of the femur and humerus occurs in patients with achondroplasia. This shortness causes the appearance of rhizomelic type of dwarfism. Since the skull of patients with achondroplasia is large during delivery, it may cause difficult delivery. During infancy, hypotonia is more dominant. Many patients also have delayed motor development.
Our aim in the study is to present the successful physical therapy and rehabilitation process of our patient diagnosed with achondroplasia for one year and her condition from diagnosis to treatment.
Our patient was born via cesarean section on 12.02.2020. The weight of the baby girl was 3500 grams. The mother's height was 162 cm. and the father's height was 174 cm. The mother was 23 years old and the father 27 years old. The same diagnosis was not present in any living relative of the patient. The patient was 1 year old when she applied to our center. She had completed head control and had not yet acquired the skills of rolling over, crawling, and sitting without support. Gross motor skills such as assisted standing and stepping were not developed. In this direction, after a 1-year physical therapy and rehabilitation period applied in 2 sessions a week, the patient gained the ability to walk independently.

Keywords: Achondroplasia, Physical therapy, Musculoskeletal diseases, Autosomal dominant

ÖZET

Akondroplazi otozomal dominant geçişli bir rahatsızlık olmasında rağmen % 80 olgu sporadikdir. Görülme sıklığı 1/25000 ile 1,5/10000 arasındadır. Akondroplazide femur ve humerusta belirgin kısalık oluşur. Bu kısalık da rizomelik tipte cücelik görüntüsüne sebep olur. Akondroplazi tanılı hastaların doğum sırasında kafatası büyük olduğundan zor doğuma neden olabilir. Bebeklik döneminde daha çok hipotoni hakimdir. Birçok hastada da motor gelişimde gecikme görülür. Çalışmadaki amacımız akondroplazi tanılı hastamızın bir yıldır devam eden başarılı fizik tedavi ve rehabilitasyon sürecini ve teşhisten tedaviye kadar olan durumunu sunmaktır.
Hastamız 12.02.2020 tarihinde sezaryen ile dünyaya gelen 3500gr. ağırlığında kız bebekti. Annenin boyu 166cm., babanın boyu ise 174 cm.'di. Anne 23, baba 27 yaşında idi. Hastanın bilinen herhangi bir akrabasında aynı tanı mevcut değildi. Hasta merkezimize başvurduğunda 1 yaşındaydı. Baş kontrolünü tamamlamış, dönme aktivitesi, emekleme ve desteksiz oturma becerilerini henüz kazanamamıştı. Destekli ayakta durma, adımlama gibi kaba motor beceriler gelişmemişti. Bu doğrultuda haftada 2 seans şeklinde uygulanan 1 yıllık fizik tedavi ve rehabilitasyon süreci sonrasında hasta bağımsız yürüme becerisini kazandı. Tedavi süreci hala devam etmektedir.

Anahtar Kelimeler: Akondroplazi, Fizyoterapi, İskelet hastalığı, Otozomal dominant

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Corresponding Author / Sorumlu Yazar: Sevda CANBAY DURMAZ, Mardin Artuklu University, Faculty of Medicine, Department of Anatomy, Mardin, Türkiye. e-mail: sevdacnby@hotmail.com

INTRODUCTION

Achondroplasia is one of the autosomal dominant skeletal diseases. A defect in the fibroblast growth factor receptor 3 (FBFR3) gene located on the fourth chromosome (4p16.3) results in achondroplasia. Although achondroplasia is an autosomal dominant disorder, 80% of cases are sporadic. The main problem is related to the development of bone tissue from cartilage. The incidence of achondroplasia is between 1/25000 and 1.5/10000 (DelRosso, Gonzalez-Toledo & Hoque, 2013).

In achondroplasia, significant shortening of the femur and humerus occurs. This shortness causes the appearance of rhizomelic type of dwarfism. Since the skull of patients with achondroplasia is large during delivery, it may cause difficult delivery. During infancy, hypotonia is more dominant. Many patients also have delayed motor development (Atabek, Aydin, Oran & Erkul 1999). Especially between 1 and 2 years of age, an increase in thoracic kyphosis and lumbar lordosis is evident in the spine. In patients with achondroplasia, walking is often delayed and develops in pathological patterns. Although thoracic kyphosis improves when the patient starts walking, lumbar lordosis may worsen (Bayhan & Mackenzie, 2014).

Patients with achondroplasia are more likely to have foramen magnum stenosis and spinal stenosis later ages compared to normal patients. For this reason, the spine should be evaluated in detail in diagnosis and treatment. Body length in achondroplasia is close to the normal value. In the lower extremities, varus deformity is more prevalent in the knees. Internal rotation of the tibia is seen with the genu varum (Şenköylü, Alanay, Zinnuroğlu, Deveci & Altun, 2011).

Sudden death due to neurological and respiratory complications can be seen in individuals with achondroplasia starting from the neonatal period. For this reason, physical therapy and rehabilitation process should be started in the early period and treatment should be continued in a multidisciplinary manner. Delay in walking in patients can accelerate musculoskeletal system problems and cause irreversible damage (Atabek, Aydin, Oran & Erkul 1999).

In this direction, we aimed to present the successful physical therapy and rehabilitation process of our patient diagnosed with achondroplasia for a year, and her condition from diagnosis to treatment.

CASE REPORT:

The study was initiated with the approval of the Ethics Committee of Inonu University Health Sciences Non-Interventional Clinical Research Ethics Committee with the 20th session dated 13-12-2022 and the decision number 2022/2999. The family was informed about the content of the study and an Informed Consent Form was signed.

Our patient was born via cesarean section on 12.02.2020. The weight of the baby girl was 3500 grams. Since the gestation period exceeded 40 weeks, the delivery was performed by cesarean section. The patient was diagnosed with achondroplasia at 32 weeks in the womb. The mother's height was 166 cm. and the father's height was 174 cm. The mother was 23 years old and the father 27 years old. No known relatives of the patient had the same diagnosis.

The patient was 1 year old when she applied to our center. She had completed head control and had not yet acquired the skills of rolling over, crawling, and sitting without support. Gross motor skills such as assisted standing and stepping were not developed. Her body awareness and body perception were quite weak, her muscle strength was quite low compared to her peers, and her tone was low. Craniofacial frontal bossing as well as enlargement around the head and midfacial hypoplasia were observed. She also had short extremities and thick blunt fingers, which are the dysmorphic features of the disease. There was no asymmetry between the extremity measurements. Although the patient did not have hip and knee flexion movements, the knees were in hyperextension. Lumbal lordosis was increased (Figure 1).

Figure 1: Patient's pretreatment condition.

During the 1-year physical therapy process of our patient, our aim is to bring the patient to an independent level in daily life activities, to maintain body smoothness. To prevent deformities that may occur, to increase muscle strength, to bring balance and coordination skills to the best level and reduce respiratory problems that are likely to develop. In addition, we aimed to keep the increase in lordosis, which is typical in achondroplasia, at a minimal level, to preserve the range of motion and to increase body awareness.

The patient underwent a physical therapy and rehabilitation program, 2 sessions a week for 45 minutes. Stretching and strengthening exercises to increase normal joint range of motion, positioning exercises to increase crawling and sitting skills, weight bearing exercises, breathing exercises, unsupported standing and walking exercises, balance and coordination exercises, and spinal stabilization exercises are included in the treatment program, progressing in line with the needs.

In this direction, after a 1-year physical therapy and rehabilitation period applied in 2 sessions a week, the patient gained the ability to walk independently (Figure 2). The treatment process is still ongoing.

Figure 2: Post-treatment status of the patient.

DISCUSSION

Achondroplasia with an incidence of 1/25000 to 1.5/10000 occurs in an autosomal dominant manner or sporadically (Akın, 2011; Bilgin et al., 2015). In our case, the absence of achondroplasia in any of their relatives, including previous generations, suggested that it may be sporadic.

Achondroplasia is the most common skeletal dysplasia characterized by short stature. The estimated height for boys is 130 cm and for girls is 120 cm. After starting to walk, deformations occur in both the knee and ankle due to the bending of the tibia (Alanay, 2011). We think that the early physical therapy and rehabilitation process had a great effect on the minimal occurrence of these deformations in our patient.

Spinal deformities and increased kyphosis and lordosis are frequently seen in patients with achondroplasia. In connection with this situation, many respiratory system problems come with it. In addition, different anomalies such as sleep disorders, snoring, partial, obstruction, central apnea or complete obstructive apnea can be seen. (Bilgin et al., 2015).

In a study, it was reported that a 12-year-old patient with achondroplasia who applied to the physician with night snoring and respiratory distress had frequent lung infections secondary to gastroesophageal reflux. The complaints of the patient who did not benefit from the CPAP treatment applied after tonsillectomy and adenoidectomy operation decreased with BIPAP, position and oxygen support treatment (Batum, Kısabay, Oğuz, Oktan, Yılmaz, 2016). There was no respiratory problem in our case. In this sense, we think that breathing exercises will be effective in minimizing the problems that may develop.

Hypothalamic-pituitary dysfunctions such as foramen magnum stenosis, brain stem compression, ventricular dilatation, and GH secretion in patients with hydrocephalus may be seen. As a result, delays may occur in motor functions (Bayhan & Mackenzie, 2014).

Our patient had retardation in motor development. Thanks to the correct and timely managed physical therapy and rehabilitation process, our patient has reached the same level as her peers.

The spine is one of the most affected areas in patients with achondroplasia. Problems such as extremity numbness, paresthesia, paraparesis and incontinence may occur as a result of degenerative changes in facet joints, intervertebral disc herniation, spondylolysis and kyphosis canal compressions (Şenköylü, Alanay, Zinnuroğlu, Deveci & Altun, 2011).

In order to prevent or minimize these situations, it is very important to provide load distribution in the body, correct posture and appropriate walking pattern.

CONCLUSION

In achondroplasia, which is diagnosed while the baby is still in the mother's womb, with the developing technology, it is of great importance to know the problems to be seen in later ages and to start the treatment process early.

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Author Contributions:

Idea/Concept: Sevda CANBAY DURMAZ, Ümran GÜVEN, Ali CANBAY, Selma SOLGUN DAĞ**; Design:** Sevda CANBAY DURMAZ, Ümran GÜVEN, Ali CANBAY, Davut ÖZBAĞ, Selma SOLGUN DAĞ**; Supervision/Consulting:** Sevda CANBAY DURMAZ, Ümran GÜVEN, Ali CANBAY, Davut ÖZBAĞ, Selma SOLGUN DAĞ**; Data Collection and/or Processing:** Sevda CANBAY DURMAZ**; Analysis and/or Interpretation:** Sevda CANBAY DURMAZ, Ümran GÜVEN, Ali CANBAY, Davut ÖZBAĞ**; Literature Review:** Sevda CANBAY DURMAZ**; Writing of the Article:** Sevda CANBAY DURMAZ**; Critical Review:** Sevda CANBAY DURMAZ, Ümran GÜVEN, Ali CANBAY, Davut ÖZBAĞ, Selma SOLGUN DAĞ

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