



Duane retraction syndrome in a patient with abnormal head position

Anormal baş pozisyonu ile gelen hastada Duane retraksiyon sendromu

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Abstract

Duane's syndrome is a rare retraction anomaly characterized by an innervation defect in the lateral rectus muscle, limitation of abduction and adduction due to the result of abnormal innervation of the horizontal rectus muscles, changes in the eyelid fissures, and abnormal vertical eye movements. The affected eye is displaced up and/or down in adduction. This syndrome, also known as Stilling-Türk-Duane syndrome, accounts for approximately 1 to 5% of all strabismus cases. In this article, we present a one-and-a-half-year-old male patient who had abnormal head position, and was diagnosed as having Duane retraction syndrome. Through this study, we want to draw attention to Duane retraction syndrome, which is one of the rare causes of strabismus.

Keywords: Abnormal head position, Duane retraction syndrome, strabismus

Öz

Duane retraksiyon sendromu lateral rektus kasında anormal sinir iletimi sonucunda gelişen abdüksiyon ve addüksiyonda kısıtlılık, yatay göz hareketlerinde kısıtlılık, göz küresinde retraksiyon, göz kapağı fissürlerinde değişiklik ve anormal dikey göz hareketleri ile belirgin ender bir kırma kusurudur. Etkilenen göz addüksiyonda yukarı ya/ya da aşağıya doğru yer değiştirir. Stilling-Türk-Duane sendromu olarak da bilinen bu sendrom tüm şaşılık olgularının yaklaşık yüzde 1 ila 5'ini oluşturmaktadır. Bu yazıda anormal baş pozisyonu nedeniyle kliniğimize başvurusunda yapılan incelemeler sonucunda Duane retraksiyon sendromu tanısı alan 1,5 yaşında bir erkek olgu sunulmuştur. Bu yazı ile ender şaşılık nedenlerinden biri olan Duane retraksiyon sendromu tanısına dikkat çekmek amaçlanmıştır.

Anahtar sözcükler: Anormal baş pozisyonu, Duane retraksiyon sendromu, şaşılık

Introduction

Duane retraction syndrome is a congenital syndrome characterized by narrowing of the lid space in adduction in the affected eye, retraction of the eyeball, and abduction limitation, frequently together with varying degrees of adduction limitation and displacement of the affected eye upwards and/or downwards in adduction. It has been reported that the most commonly accepted cause of Duane retraction syndrome is congenital structural anomalies (1). Its prevalence is 1–5% among the causes of strabismus and 0.1% in the general population (2). Patients with Duane retraction syndrome may exhibit

strabismus (esophoria or exotropia). The syndrome is generally classified in three different types. Treatment of Duane retraction syndrome is similar to the treatment of the other types of strabismus. In this article, we discuss a 1.5-year-old male patient who presented to our clinic with abnormal head position and was diagnosed as having Duane retraction syndrome type 1.

Case

Our patient was a 1.5-year-old boy. He presented to our clinic with symptoms of strabismus in his left eye and abnormal head position. His symptoms began about six months ago and had increased gradually. His personal

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and family history revealed no pathology. At the time of hospitalization, his weight was 11 kg (25–50p), his height was 67 cm (25p), and his head circumference was 47 cm (25p). His vital signs were found to be normal. On systemic examination, his neck was deviated towards the right side, his chin was deviated towards the left side and upwards in the primary gaze position. As a result of an ophthalmologic consultation, it was stated that monocular object and light tracking was good, gazes in all directions were normal in the right eye, and no refractive error was found in either eye. Anterior and posterior segment findings were found to be natural and limitation of abduction was present in the left eye. Marked esophoria was absent in the primary gaze position (Fig. 1, 2). Other system examinations were found to be normal.

Duane retraction syndrome type 1 was primarily considered with the abnormal head position and limitation of abduction in the left eye. The patient was followed up by the strabismus unit. Blood tests results were as follows: white blood cells: 9440/mm³, hemoglobin: 12.1 g/dL, platelet count: 262,000/mm³, C-reactive protein: 0.1 mg/dL, liver and renal function tests: normal. Hearing screening was performed in terms of potential extraocular findings that could accompany, which was found to be normal. Direct cervical radiography revealed no anomalies. Cranial magnetic resonance imaging (MRI), MRI angiography and venography were found to be normal. Investigations performed in terms of cardiac and renal anomalies revealed no pathology. The patient was given outpatient follow-up visit appointments. Verbal consent was obtained from the patient's parents.

Discussion

Duane retraction syndrome is a congenital anomaly that is mostly sporadic and also has autosomal dominant inheritance (DURS1, 8q13, 2q31). It was described by Stilling and Türk for the first time at the end of the 19th century, and was subsequently named Duane retraction syndrome when Duane described the clinical picture in 1905. This congenital anomaly leads to failure in the normal development of the abducens nerve and abnormal nerve conduction in the lateral rectus muscle by the ocular motor nerve. As a result of these pathologies, retraction in the bulbus oculi and narrowing of the palpebral fissure occur in adduction. This condition occurs because of simultaneous contraction of the medial and lateral rectus muscles during attempted adduction (3). Duane retraction syndrome constitutes 1–5% of patients who are followed up for strabismus, and is observed with a rate of 0.1% in the population, more frequently in women (4).

Esophoria or exotropia may be observed in patients with



Figure 1. Abnormal head position in the primary gaze position



Figure 2. Marked limitation of abduction in the left eye during attempted lateral gaze

Duane retraction syndrome and the disease is classified in three groups according to these findings. In type 1 Duane retraction syndrome, marked limitation in abduction is present in the affected eye and this type constitutes 70–80% of cases. Limitation of adduction is present in type 2 and both adduction and abduction are limited in type 3 (4). The left eye is affected in more than half of all cases of Duane retraction syndrome. Some patients may appear normal in the primary gaze position and pathology can only be detected in the lateral gaze position (5). Other clinical characteristics include fusion of the affected eye in adduction and abnormal leaning or rotation of the head (having abnormal head position). It is thought that upward movement or downward movement

of the affected eye during fusion arises from abnormal nerve conduction of the vertical rectus muscles. In the study conducted by Zhang et al. (6) with 201 patients, abnormal head position was observed with a rate of 20.9%. The most common accompanying ocular abnormality is nystagmus in Duane retraction syndrome. In addition, epibulbar dermoid, anisocoria, and ptosis may also be observed in these patients. In addition, Marcus-Gunn jaw-winking syndrome, congenital cataract, optic nerve hypoplasia, and heterochromia have also been reported in the literature. Amblyopia and other refractive errors (anisometropia) may also be observed in patients with Duane retraction syndrome (7). Our patient had an abnormal head position and presented to our clinic because of this symptom.

In Duane retraction syndrome, extraocular findings including deafness, asymmetrical face, cleft palate, outer ear anomalies, preauricular tag, vertebral anomalies (fusion of the second and third cervical vertebrae), thenar hypoplasia, rib and foot anomalies, and cardiac anomalies, and association with Goldenhar syndrome and Klippel-Feil syndrome, Wildervanck syndrome, and Duane-radial ray syndrome have been reported. Rarely, renal dysplasia, vesicoureteral reflux, Hirschsprung disease, segmental dilatation of the colon, focal segmental glomerulonephritis, imperforated anus, umbilical hernia, genitourinary anomalies and Holt-Oram, Moebius, and Marfan-Ehler Danlos syndromes may be observed in association with Duane retraction syndrome (5, 8). When our patient was evaluated in terms of extraocular involvement in our clinic, no additional anomalies were found in the physical examination. The hearing test was found to be normal. Electrocardiogram and echocardiogram performed in terms of probable cardiac pathologies were found to be normal. Direct cervical and vertebral radiographies obtained in terms of Klippel-Feil anomaly were found to be normal. A fundoscopic examination performed in relation with Wildervanck syndrome, and cranial MRI performed in terms of probable intracranial space occupying lesions and extraocular muscle anomalies revealed no pathology. Treatment of Duane retraction syndrome is similar to the other strabismus therapies. Surgical options may be preferred in the presence of abnormal head position and marked enophthalmos in adduction. However, most patients do not have very severe head position and they have normal vision in both eyes. Therefore, surgical treatment is not necessary at the first stage. Surgical treatment is performed in patients who are symptomatic because of marked strabismus (9). In our patient, no surgery was planned by our ophthalmology unit because no severe internal strabismus was present and the abnormal head position was not severe. We started to follow

up the patient at outpatient visits. In conclusion, pediatric patients who present with strabismus should be evaluated by ophthalmologists and pediatricians in association. In the presence of strabismus and abnormal head position, the diagnosis of Duane retraction syndrome should be considered and patients should be examined in detail in terms of probable additional accompanying anomalies.

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