HORIZONTAL GAZE PALSY AND SCOLIOSIS

A Case Report

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ABSTRACT

Horizantal gaze palsy (HGP) in association with scoliosis has been reported in 42 patients both in orthopaedic and ophtalmologic literature. Bilateral congenital HGP is a rare disorder and the etiology of the gaze and associated scoliosis remains unclear. A maldevelopment or severe dysfunction of neurons of the paramedian pontine reticular formation (PPRF) was supposed by many researcher to be the causative lesion of HGP. Here we report a sporadic case with scoliosis and horizantal gaze palsy.

Key words: Horizantal gaze palsy, Scoliosis, Convercence

ÖZET

HORİZONTAL BAKIŞ KISITLILIĞI VE SKOLYOZ

Skolyoz ile Horizontal Bakış Kısıtlılığı (HBK) birlikteliği ortopedik ve oftalmolojik literatürde 42 hasta sunumu ile bahsedilmektedir. Çift taraflı konjenital HBK nadir bir sendromdur ve bakış kısıtlılığı ile skolyozun etyolojisi aydınlatılmamıştır. HBK'nın etyolojisinde paramedian pontin retiküler formasyon ciddi disfonksiyonu birçok araştırıcı tarafından önerilmektedir. Bu yazıda skolyoz ve HBK olan sporadik hasta sunmaktayız.

Anahtar sözcükler : Horizontal bakış kısıtlılığı, Skolyoz, Konverjans

INTRODUCTION

Horizontal gaze palsy (HGP) in association with scoliosis has been reported in 42 patients both in orthopaedic and ophtalmologic literature (1,16). Juvenile progressive scoliosis in combination with congenital horizontal gaze palsy apparently is caused by a malfunction of the normal control mechanism for equilibrium related to the lower brainstem. Bilateral congenital HGP is a rare disorder and the etiology of the gaze palsy and associated scoliosis remains unclear. A maldevelopment or severe dysfunction of neurons of the paramedian pontine reticular formation (PPRF) was supposed by many researcher to be the causative lesion of HGP. Bilateral HGP may be isolated (17,18) or associated with other syndromes such as Klippel-Feil syndrome (19), Fascial hemiatrophy (20), Moebius syndrome (4), Ear dysplasia (Goldenhar-Gorlin syndrome) (21), Situs inversus of the optical disc (20), congenital scoliosis (22). A common pathogenesis for these three anomalies not previously associated is not apparent. The relationship between neurologic diseases and adolescent idiopathic scoliosis has been well recognized and shown to be related with postural disequilibrium and abnormality of the vestibular functions, defective proprioceptive functions and ocular motor disturbances (23,24).

CASE REPORT

A 19 year-old girl had spinal deformity since 16 and received no treatment. First degree consanguinity was present. She is the third of four brothers and sisters. There were two intrauterine deaths without any diagnosis evident to the mother. She was seen at the department of orthopaedics due to progressive deformity and reffered to department of ophtalmology because of abnormal horizontal eye movements. On

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examination, her visual acuity was 6/12 in both eyes. She was orthoptic; there was absence of all conjugate horizontal eye movements (Figure 1).

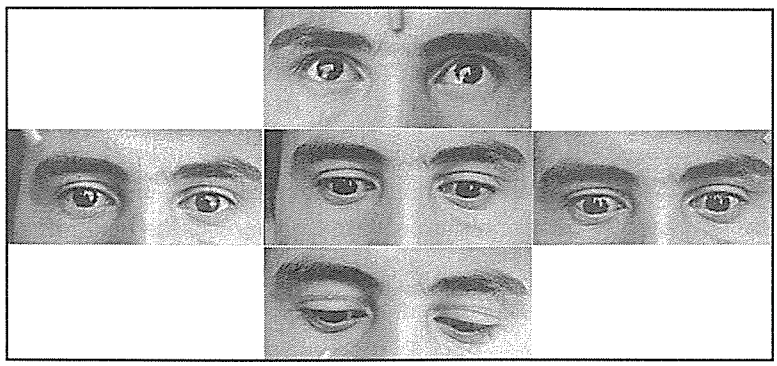

Figure 1. Horizontal paralysis is evident on 9 cardinal positions.

Her vertical eye movements and convergence were normal. She has had head nodding and marked horizontal nystagmus since birth. The pupils were equal and reacted normal. Anterior and posterior segments were normal. Physical examination revealed a right thoracic scoliosis with imbalance, head nodding, and a normal neurologic examination. She has had her menses three years ago. No hearing loss was detected. Cranial and spinal MRI revealed no abnormality. Preoperative x-ray evaluation revealed a left 2 cm. trunk shift and 2 cm. left shoulder imbalance. Cobb angle measurements were T5-L1 60° and L1-L5 40°. Due to documented progression of her spinal deformity she was operated. At first stage anterior diskectomy from T8 to L2, two weeks later posterior instrumented correction and fusion was performed using Alıcı spinal system (İzmir, Hipokrat A.Ş.). Immediate postoperative measurements revealed 28° thoracic and 17° lumbar residual curvatures with (-2 mm) balanced spine. Shoulder imbalance was reduced to 7 mm. Two years postoperatively thoracic and lumbar Cobb angle measurements were 28° and 17° respectively (Figure 2). Left shoulder was 2 cm. elevated as in preoperative evaluation. She was happy with the body image at the last follow-up.

DISCUSSION

The eye movements of the present cases showed a common pattern where no horizontal eye movements could be elicited voluntarily or by visual, vestibular stimuli but convergence and vertical movements were possible. Consequently diagnosis of bilateral HGP can be given. Review of the ophtalmologic and orthopedic literature shows reports of congenital HGP that can present as a single symptom or in combination with other syndromes (1-16). In 1970, Dretakis (3) first reported three cases of familia idiopathic scoliosis with congenital HGP. In literature, the mean age of diagnosis for HGP was 2.8 years (range: 2 weeks to 21 years) while for scoliosis it was 4 years (6 months to 12 years). During follow-up, ocular findings were stable; however scoliosis was progressive in almost all cases. Horizontal nystagmus was accompanied 70% of cases.

Juvenile progressive scoliosis in combination with congenital familiar HGP apparently is caused by a malfunction of the normal control mechanism for equilibrium related to the lower brain stem.

A gaze center for horizontal eye movements has been postulated to exist in the vicinity of the abducens nucleus within the pons. The region that is believed to be responsible for generating commands for conjugate horizontal eye movement is a part of the trochlear nuclei. It is usually referred as the paramedian pontine reticular formation (PPRF). The afferent connections of the PPRF include projections from the vestibular nuclei, from cerebellum, from the superior colliculus. Other regions that may send projections to the PPRF are the frontal eye fields, the pretectal nuclei, the inferior colliculus, the spinal cord and perihypoglossal nuclei (25,26).

The frontal eye fields and superior colliculus are involved in the production of saccades, while the parieto-occipital-temporal junction region is thought to be important in the production of pursuit. Axons from the cell located at PPRF travel to the ipsilateral

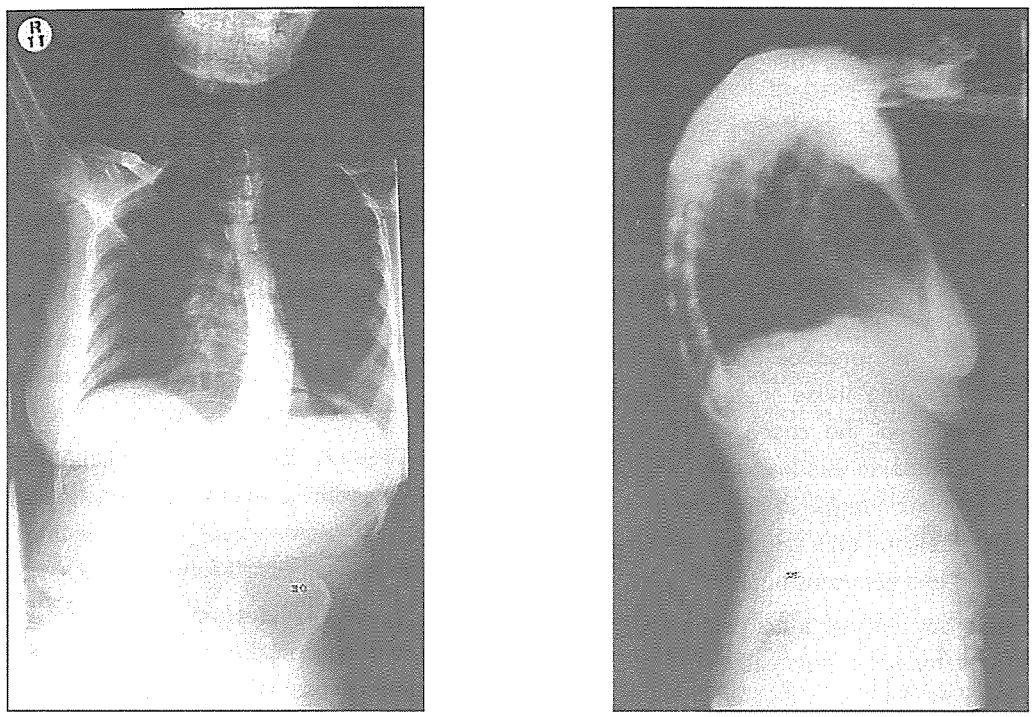

Figure 2.a,b. Preoperative X-rays demonstrate trunk imbalance.

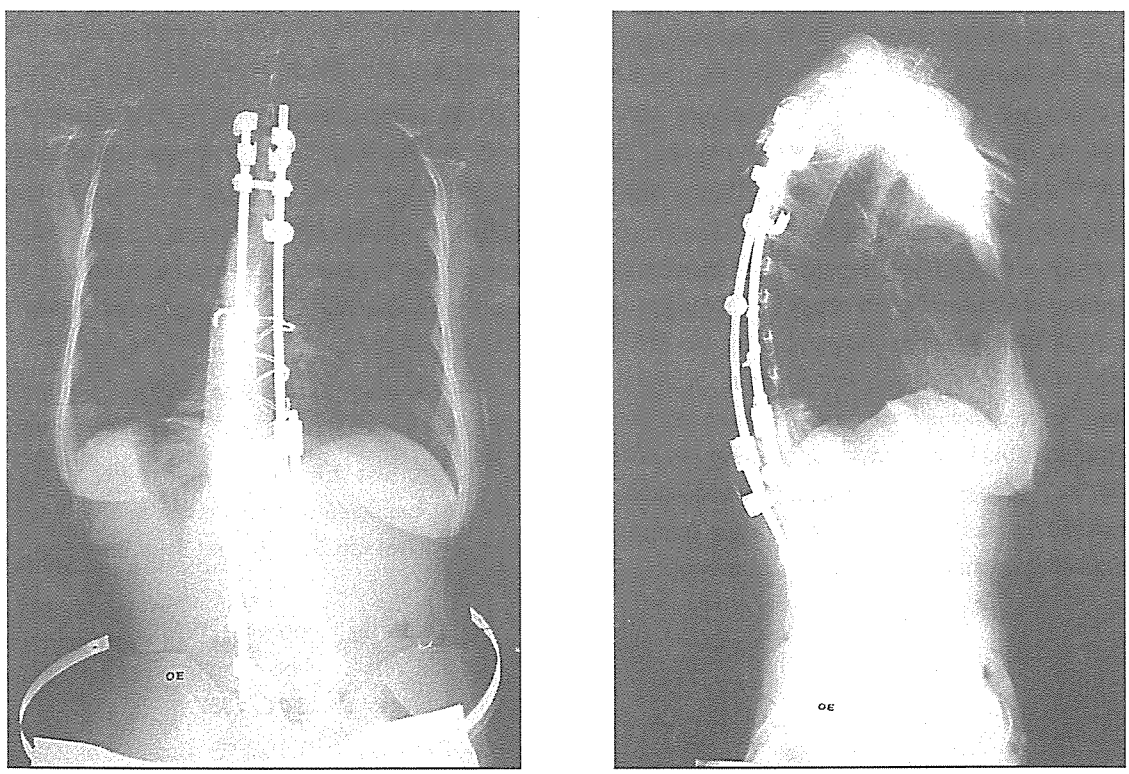

Figure 2.c,d. Postoperative AP and Sagittal views demonstrate an acceptable correction of both planes.

abducens nucleus where they synapse with abducens motorneurons whose axons travel to the ipsilateral lateral rectus muscle and abducens internuclear neurons whose axons cross the midline and travel in the medial longitudinal fasciculus (25,26).

A maldevelopment or a severe dysfunction of neurons of the PPRF was supposed by many researches to be the causative lesion (1,25,27). Several other reports have suggested that bilateral lesions of PPRF can produce loss of all rapid eye movements, including those in the vertical plane. However, in none of the cases, restriction of the vertical eye movements was found and it is difficult to consider that the main region to cause HGP was in the PPRF. It is possible that anomaly is somewhere between PPRF and abducens nuclei.

It is demonstrated that a brainstem lesion at the level of the lateral nucleus vestibular is, nucleus gracilis or the colliculus superiors could induce spinal deformities in rats (28,29). One-half of the patients with idiopathic scoliosis demonstrated dysfunction of the brainstem center which controls the postural regulation system. It seems probable that HGP associated with progressive scoliosis form a separate clinical entity and causative lesion needs further research.

Several familial cases with the clinical combination of lateral gaze palsy and progressive scoliosis have suggested an autosomal recessive inheritance triad (25). This triad was confirmed also by the parental consanguinity as reported in 15 cases in literature review. As a conclusion every child with congenital HGP should be evaluated for a possible associated scoliosis. If present, a diagnosis of this presumably autosomal recessive syndrome can be made with appropriate treatment and genetic counseling.

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