

## Brain MRI Features in Horizontal Gaze Palsy with Progressive Scoliosis

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**Received:** February 04, 2017

**Accepted:** April 27, 2017

**Published:** April 28, 2017

**Citation:** Almasi M, Rohani M, Soltan Sanjari M, Imani A. 2017. Brain MRI Features in Horizontal Gaze Palsy with Progressive Scoliosis. *J Neuroimaging Psychiatry Neurol* 2(1): 1-2.

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### Abstract

Horizontal gaze palsy with progressive scoliosis (HGPPS) is a rare disease with characteristic imaging features. We report a teenager girl with severe kyphoscoliosis and bilateral horizontal gaze palsy with normal other neurologic examination. The brain MRI revealed “tented” pons and “butterfly-shape” medulla. These changes are due to agenesis of deep structures of brainstem which play important roles in horizontal gaze and proprioception.

### Keywords

Horizontal gaze palsy, Progressive scoliosis, MRI features

### Case Report

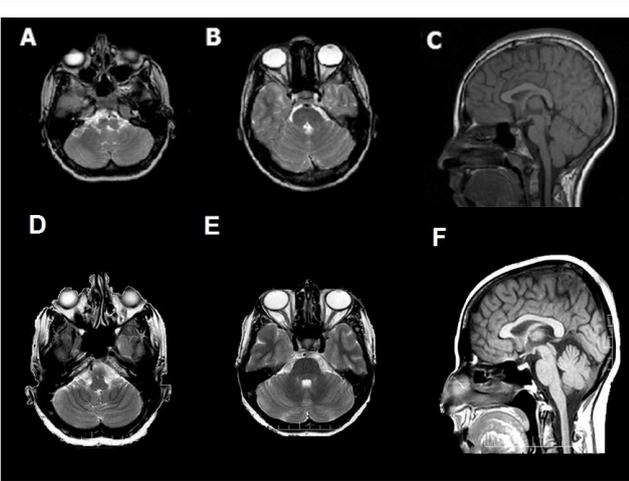
A 17-years old girl referred to our hospital for repair of scoliosis. The scoliosis began since 2 years ago with a progressive course and created much disability in patient’s daily life. Beside severe kyphoscoliosis (Figure 1), she had bilateral horizontal gaze palsy, but the other neurologic examinations including facial expression, pupil size and reaction, convergence, vertical gaze and other eye movements were normal (Figure 2). It was not possible to overcome to limitation of horizontal gaze with vestibuloocular maneuver. She had no birth difficulty or developmental delay and her intelligence and school performance were intact. Her sister had also scoliosis in her teens but the family denied any other neurologic problem.



**Figure 1:** Antero posterior view of X-ray shows severe scoliosis.



**Figure 2:** Examination of eye movements; which discloses bilateral horizontal gaze palsy with normal vertical gaze.



**Figure 3:** Axial T2 and Sagittal T1-weighted MRI of the patient, atrophy of medulla with a butterfly-shape (A); dorsal midline cleft and tented pons (B); and absence of facial colliculi (C). Axial T2 and Sagittal T1-weighted MRI of a normal subject at the similar anatomic levels, including the medulla (D), the pons (E) and the sagittal midline of brainstem (F).

The brain MRI showed significant pontine and medullary atrophy with “tented” pons and “butterfly-shape” medulla

appearances (Figure 3A and 3B). Absence of facial colliculi in the floor of fourth ventricle are prominent at the sagittal plan of brain stem (Figure 3C). Based on clinical features and typical brain imaging features, the diagnosis of “Horizontal gaze palsy with progressive scoliosis” (HGPPS) was made; although unfortunately the patient lost the follow-up without any future investigations, including genetic study.

Horizontal gaze palsy with progressive scoliosis (HGPPS) is a rare disease with bilateral horizontal gaze palsy and severe scoliosis and characteristic imaging features [1]. The absence of facial colliculi which is an anatomic structure at the floor of fourth ventricle is due to selective agenesis of abducens nuclei [2] and gives rise to the picture of “a tent” to pons. In addition to abducens nuclei, maldevelopment of medial longitudinal fasciculus may lead to a dorsal midline cleft along the pons [2, 3]. Also there is a ventral midline medullary cleft which is thought to be as the result of uncrossed corticospinal fibers [4]. The gracilis and cuneatus nuclei and medial lemniscus are absent in HGPPS and lead to a prominent decrease in the size of medulla oblongata [3]. All of these structures are a part of proprioceptive systems that can explain the scoliosis.

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