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Horizontal Gaze Palsy Associated with Progressive Scoliosis: Magnetic Resonance Imaging Findings

İlerleyici Skolyoz ile Birlikte Horizontal Bakış Felci: Manyetik Rezonans Görüntüleme Bulguları

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Dear editor,

A six-year-old girl presented with visual impairment in both eyes. Her family history was negative for consanguineous marriage. On physical examination, there were no horizontal eye movements. Vertical gaze and convergence were normal in both eyes, and both pupils were equal in size. The patient had global growth retardation, and levoscoliosis of the thoracolumbar vertebrae was noticed in an anteroposterior plain radiogram (Figure 1). Other neurological findings and routine laboratory studies were normal. Brain magnetic resonance imaging (MRI) showed hypoplasia of the medulla oblongata and pons, a deep midline pontine cleft starting from the floor of the fourth ventricle and extending anteriorly, flattening and tenting of the floor of the fourth ventricle at the level of the pons, bilateral absence of facial colliculi, and the butterfly sign in the medulla oblongata (Figure 2A, B, C, D). Diffusion tensor imaging (DTI) showed an absent crossing of the superior cerebellar peduncle fibers at the level of the pons and anterior displacement of the transverse pontine fibers (Figure 2E). The patient was diagnosed with horizontal gaze palsy with progressive scoliosis (HGPPS). The family members were offered a *ROBO3* gene test, but they refused it. Written consent was obtained from the patient's relatives.

HGPPS is a rare congenital anomaly with autosomal recessive inheritance caused by a mutation of the *ROBO3* gene on chromosome 11q23-35, which affects the main axonal tracts such as the corticospinal and dorsal column–medial lemniscus tracts

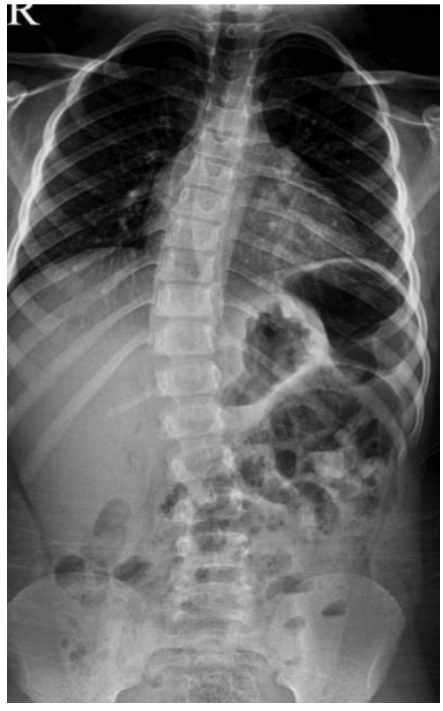

Figure 1. Anteroposterior spine radiography demonstrates thoracolumbar scoliosis, showing convexity toward the right

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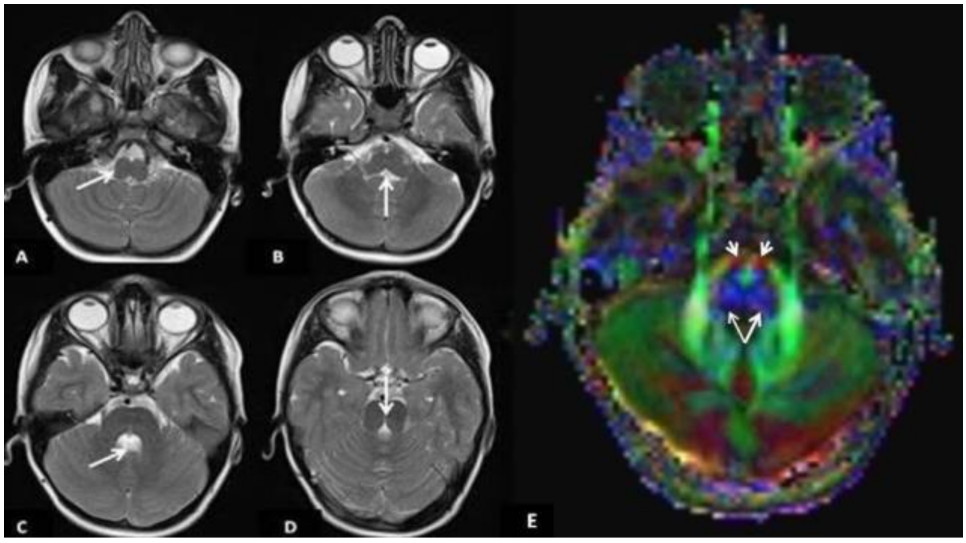

Figure 2. T2-weighted axial magnetic resonance images show the butterfly appearance of the medulla oblongata (A, arrow), flattening and tenting of the floor of the fourth ventricle (B, C; arrows), and a deep midline pontine cleft starting from the floor of the fourth ventricle and extending anteriorly (D, arrow). (E) The color-coded fractional anisotropy map on the diffusion tensor imaging shows an absence of the decussation of the superior cerebellar peduncles, anterior displacement of transverse pontine fibers (small white arrows), and shrinking of the afferent fibers of the inferior cerebellar peduncles (arrows)

and manifests with kyphoscoliosis and abnormal horizontal gaze. Under normal conditions, horizontal gaze requires the activation of the ipsilateral abducens nerve and contralateral oculomotor nerve and is mediated by the nucleus abducens. The latter is located on the lateral part of the tegmentum on the floor of the fourth ventricle and surrounded by the facial nerve fibers. Facial colliculus is seen as a pair of elevations on the floor of the fourth ventricle. An absent facial colliculus indicates nervus abducens agenesis in the absence of conjugated eye movements. In HGPPS, the congenital absence of the abducens nerve nucleus results in an abnormal horizontal gaze. The etiology of progressive scoliosis in patients with HGPPS is not entirely clear. However, it is thought to primarily result from neurological dysfunction of the proprioceptive nerves found in the medial lemniscus and the posterior column of the spinal cord. Scoliosis is a more important sign for HGPPS than the ophthalmological signs (1,2,3,4).

MRI findings in HGPPS include the loss of facial colliculus due to the absence of the abducens nerve, deep split pons sign, butterfly medulla oblongata, and tenting of the fourth ventricle. The deep split pons sign, clearly visualized in the axial plane T2-weighted MRI series, is key to distinguishing HGPPS from other differential diagnoses (2,3,4,5). DTI findings have not been completely defined in cases of HGPSS, and various signs have been found in different studies. In DTI studies, multiple signs, including the absence of the crossing of the superior cerebellar peduncular fibers, shrinking of the afferent fibers of the inferior cerebellar peduncles, and anterior displacement of the transverse pontine, were identified (5).

Differential diagnosis of HGPSS should include Moebius syndrome and Duane retraction syndrome type III (3,4).

HGPPS should be considered in the differential diagnosis of cases of bilateral horizontal gaze palsy. MRI is the most important diagnostic tool used in conjunction with typical signs of the disease.

Ethics

Informed Consent: Obtained.

Peer-review: Externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: M.H.A., Concept: M.H.A., Ö.K.Y., N.B., Design: M.H.A., Ö.K.Y., N.B., Data Collection or Processing: M.H.A., N.B., Analysis or Interpretation: M.H.A., Ö.K.Y., N.B., Literature Search: M.H.A., Ö.K.Y., N.B., Writing: M.H.A., N.B.

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