MR Imaging of Brainstem Hypoplasia in Horizontal Gaze Palsy with Scoliosis

Rossi, A.¹· Catala, M.²· Biancheri, R.¹· Di Comite, R.¹· Tortori-Donati, P.¹
¹G. Gaslini Children’s Research Hospital, Genova, ITALY, ²Pitié-Salpêtrière Hospital Group, Paris, FRANCE.

Purpose
To present brain MR imaging findings in a patient with horizontal gaze palsy with scoliosis (HGPS), and to discuss the pathogenesis and possible embryologic substratum of this rare entity.

Materials & Methods
A 13-year-old girl with early onset thoracolumbar scoliosis was admitted to our institution to undergo surgical correction of her spinal deformity. She also had congenital, complete lack of horizontal eye movements with substantial preservation of vertical gaze. She compensated for her deficit by turning her head in the desired direction, thereby obtaining regular binocular vision. Neurologic examination was otherwise normal without cognitive abnormality. The girl underwent brain MR imaging prior to spinal surgery.

Results
MR imaging revealed a hypoplastic pons whose posterior two thirds were split into two halves by a midsagittal cleft extending ventrally from the fourth ventricular floor. The prominence normally produced by the abducens nuclei on the fourth ventricular floor was absent. The medulla was also hypoplastic and showed a butterfly configuration. There also was a thickened corpus callosum.
Conclusion

HGPS is a rare autosomal recessive disorder characterized by congenital absence of conjugate horizontal eye movement with progressive scoliosis developing in childhood or adolescence. Horizontal gaze palsy has been related to dysfunction of abducens nuclei and medial longitudinal fasciculus (MLF), whereas scoliosis is thought to be caused by lower brainstem abnormalities involving the dorsal longitudinal fasciculus (DLF). Brain MR studies have been reported only exceptionally, and have either been normal or displayed brainstem hypoplasia. The pons is developed from the ventrolateral wall of the metencephalon approximately between gestational weeks 5 and 8. During this period, the developing fourth ventricle shows a ventral furrow that deeply indents the posterior aspect of the metencephalon. Failed development of medial pontine structures including the abducens nuclei and MLF could result into persistence of an abnormally deep ventral fourth ventricular furrow. A butterfly configuration of the medulla is further evidence of maldevelopment of dorsomedial structures, possibly including the DLF. Since no cognitive disturbances were recorded, the significance of callosal thickening remained undetermined. Our results provide support for the theory that maldevelopment of dorsal brainstem structures plays a crucial role in the pathogenesis of HGPS. MR imaging can reveal structural brainstem abnormalities in these patients.

References