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Congenital Horizontal Gaze Palsy, Progressive Scoliosis
Brain MRI in a 13 year old girl with HGPPS revealed:

1. A hypoplastic pons in which the posterior two-thirds were split into two halves by a midsagittal cleft extending ventrally from the fourth ventricular floor, generating a split pons sign on axial images.

2. The facial colliculi were absent, and the fourth ventricular floor was tent shaped.

3. The medulla was also hypoplastic and showed a butterfly configuration.

4. The inferior olivary nuclei were prominent with respect to the pyramids, and the prominence of the gracile and cuneate nuclei on the posterior aspect of the medulla was absent.
Figure 1. MR images obtained in a 13-year-old girl with early-onset thoracolumbar scoliosis.

A. Sagittal T1-weighted image (500/12(TR/TE)) of the brain shows depression of the floor of the fourth ventricle (arrowhead). The pons and medulla oblongata have a reduced volume.
Figure 1B. Axial T2-weighted image (4500/120) at the level of the medulla oblongata shows rectangular configuration of the medulla. The floor of the fourth ventricle is tent shaped (arrows), with missing prominence of the cuneate and gracile nuclei. The inferior olivary nuclei (IO) are prominent with respect to the pyramids (P).
Figure 1C. Axial T2-weighted image (4500/120) at the level of the pons shows absence of the facial colliculi, with tent shaped configuration of the floor of the fourth ventricle (arrows). A deep midsagittal cleft extends ventrally from the fourth ventricular floor, producing the split pons sign (arrowhead).
Figure 2. MRI of the spine showing prominent scoliosis

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Congenital Horizontal Gaze Palsy with Progressive Scoliosis (HGPPS)

Mutation ROBO 3 Gene/Chromosome 11q23-q25

Congenital Cranial Disinnervation Syndrome
Eye Movements

Horizontal Gaze Palsy

Horizontal Vestibular Ocular Reflex Absent

Esotropia

Preservation of Convergence

Normal Vertical Gaze

Head Saccades
Etiology

HGPPS is one of several genetic disorders of eye and lid control that are believed to result from cranial nuclear maldevelopment.

Among these entities, the most closely related to HGPPS are:
1. Duane retraction syndrome and
2. Mobius syndrome.

Maldevelopment of the abducens nucleus plays a crucial role in the pathogenesis of both these entities, as well as of HGPPS.
Congenital Cranial Disinnervation Syndrome

Features:
- Present at birth
- Usually non-progressive
- Have an \textit{autosomal inheritance} pattern, that may occur sporadically
- May result from \textit{primary disinnervation}, i.e. failed or misguided development of neurons or from \textit{aberrant innervation} during development (i.e. \textit{secondary disinnervation}).


Acknowledgement

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