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and Scoliosis**

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CASE REPORT

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MR Imaging Features of Brain Stem Hypoplasia in Familial Horizontal Gaze Palsy and Scoliosis

SUMMARY: We report the case of a child with horizontal gaze palsy, pendular nystagmus, and discrete thoracolumbar scoliosis. MR imaging of the brain depicted pons hypoplasia with an absence of the facial colliculi, hypoplasia, butterfly configuration of the medulla, and the presence of a deep midline pontine cleft (split pons sign). These MR imaging findings suggest familial horizontal gaze palsy with progressive kyphoscoliosis, a rare congenital disorder. To the best of our knowledge, MR imaging findings of only 4 similar cases, with or without progressive idiopathic scoliosis, have been reported. We discuss the pathogenesis substratum of this entity. Early recognition of this rare entity is important if supportive therapeutic measures in progressive scoliosis are to be applied.

Isolated malformations involving the pons and medulla oblongata are extremely rare. They usually occur associated with disorders of the cerebellum. Horizontal gaze palsy with progressive scoliosis (HGPPS) is a rare congenital disorder with autosomal recessive inheritance, believed to result from cranial nuclear maldevelopment and characterized by absence of conjugate horizontal eye movements, preservation of vertical gaze and convergence, progressive scoliosis developing in childhood and adolescence, midline pontine cleft, butterfly configuration of the medulla, brain stem hypoplasia, and absence of facial colliculi.¹ Although several clinical cases of brain stem hypoplasia with familial HGPPS have been published, Pieh et al and Rossi et al were the only authors to report on MR imaging findings of HGPPS.^{2,3} Congenital cleavage of the dorsal pons and medulla oblongata associated with horizontal gaze palsy without scoliosis in 2 children have also been reported.^{4,5}

We report the clinical and imaging findings of one child with horizontal gaze palsy and congenital midsagittal cleft in the dorsal pons, discuss the pathogenesis of this rare entity, and hypothesize that early recognition of these clinical and imaging findings could help prevent severe scoliosis by enabling application of supportive therapeutic measures at an early phase of the disease.

Case Report

A 20-month-old boy born at term to healthy nonconsanguineous parents presented with hypotonia and abnormal eye movements since the neonatal period. Neurologic and ophthalmologic examinations revealed pendular nystagmus and total absence of horizontal gaze consistent with involvement of the abducens nucleus, without pseudobulbar or long tracts signs. Convergence and pupillary reflexes were normal. Discrete thoracolumbar scoliosis and slight mental retardation were also present. MR imaging findings revealed a hypoplastic pons with a midsagittal cleft extending ventrally from the fourth ventricular floor, generating a split axial pons sign on axial images and hypoplasia of the medulla oblongata with butterfly configuration (Figs 1 and 2). The fourth ventricular floor was tent shaped, and the facial colliculi were absent (Fig 3). The cerebral hemispheres, corpus callosum, and cerebellum were normal (Fig 4).

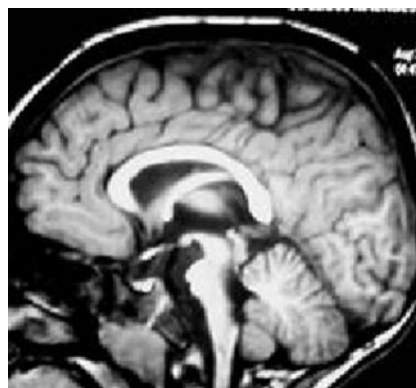


Fig 1. Sagittal T1-weighted image. Hypoplasia of the pons and medulla oblongata. Normal corpus callosum.

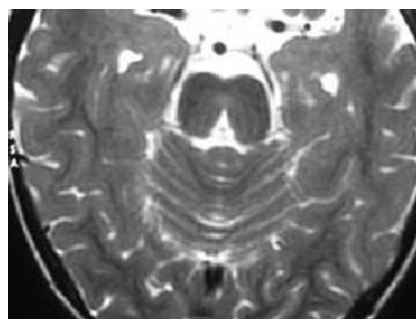


Fig 2. Axial T2-weighted image. Deep midsagittal cleft extends ventrally from the fourth ventricular floor originating the split pons sign.

Discussion

Smooth horizontal gaze is a coordinated activity controlled by the abducens nucleus. This nucleus, located in the lower part of the pontine tegmentum at the level of the fourth ventricular floor, contains 2 types of neurons, one directly innervating the ipsilateral lateral rectus muscle and the other consisting of internuclear neurons that project through the medial longitudinal fasciculus (MLF) to the contralateral oculomotor nucleus, promoting the coordination of eye movement.

Facial colliculi, a paired prominence at the floor of the fourth ventricle, result from axons of the motor nuclei of the facial nerves passing dorsally to the abducens nuclei.

Absence of conjugate horizontal eye movement and facial colliculi not visible on axial MR imaging with intact facial, vestibulocochlear, and oculomotor nerves suggest selective agenesis of the abducens nuclei.

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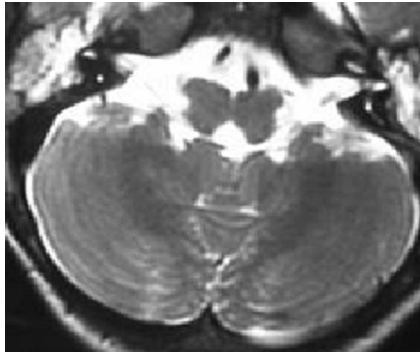


Fig 3. Axial T2-weighted image. Hypoplasia of the medulla oblongata with butterfly morphology.

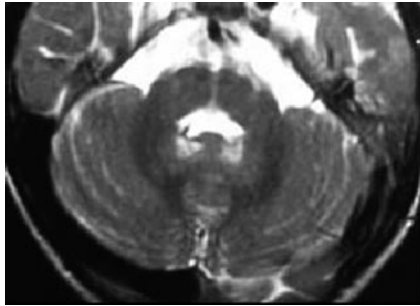


Fig 4. Axial T2-weighted image. At the level of the pons, the floor of the fourth ventricle is flattened and has a tent shape. Absence of the facial colliculi is notable.

The split pons sign found on the axial MR imaging was similar to those previously reported by Rossi et al, Squirrel et al and by Nuri Sener.³⁻⁵ This could be due to the abnormal development of the abducens nuclei and medial longitudinal fasciculus occurring between gestational weeks 5 and 8 as the ventral furrow that indents the posterior metencephalon progressively disappears while the dorsomedial nuclei and tracts develop.

Progressive idiopathic scoliosis has been described in childhood. Although its pathogenesis is still poorly understood, a primary neurologic dysfunction involving the proprioceptive inputs mediated by the posterior column pathways of the spinal cord and medial lemniscus, the postural equilibrium, and the labyrinthine function mediated by the vestibular nuclei and the interplay of visual and vestibular reflexes mediated by the vestibular nuclei and the medial longitudinal fasciculus have been described as a possible explanation.³ In our case, the discrete scoliosis could be explained by the child's age or by this being a genetic syndrome with variable expression.

Conditions such as Möbius syndrome and Duane retraction syndrome have similar clinical findings and should be included in the differential diagnosis. Abnormal development of the abducens nucleus plays a crucial role in the pathogenesis of both of these entities.

Möbius syndrome, a congenital disorder characterized by partial or complete facial diplegia and horizontal gaze palsy, with frequent involvement of other cranial nerves, results from agenesis or destruction of both the abducens and facial nuclei, possibly accompanied by abnormalities of other cranial nerve nuclei.⁵ Duane retraction syndrome is characterized by absent abduction, insufficient adduction, globe retraction, and palpebral narrowing during fixation,⁶ caused by selective absence of direct abducens nerve motoneurons with preservation of internuclear neurons visible.¹ Aplasia of abducens nerve in Duane retraction syndrome and hypoplasia of both abducens and facial nuclei associated with hypoplasia of the caudal of the brain stem and absence of hypoglossal eminence in Möbius syndrome have been rarely reported as some of the imaging features of Duane retraction and Möbius syndromes.^{5,7} There are no reports of split pons sign associated with these syndromes. If this is the case, it can also help in the differential diagnosis of genetic eye motility disorders.

Conclusion

MR imaging may play a critical role on the precocious diagnosis of HGPPS by showing the maldevelopment of the dorso-medial brain stem structures and absence of the facial colliculi, believed to result from agenesis of the abducens nerves.

Early diagnosis of this congenital entity may be important to apply supportive therapies to prevent rapid progression of idiopathic scoliosis.

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