Rhombencephalosynapsis and Chiari II Malformation with Spinal Deformities

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Abstract

Rhombencephalosynapsis is a rare congenital intracranial malformation. Partial rhombencephalosynapsis is a variation of this anomaly with few reported cases. We report a unique case of a patient with partial rhombencephalosynapsis associated with Chiari II, and various spinal malformations, which is very rare, and only few such cases have been reported in literature. These findings suggest that rhombencephalosynapsis can be associated with spinal malformations and, furthermore, that cases with the common features of rhombencephalosynapsis and Chiari II malformation can exist. Such an association likely represents a new anomaly of the hind brain with spine.

Keywords: Blocked vertebrae, Chiari II malformation, Kyphoscoliosis, Rhombencephalosynapsis, Split cord.


Introduction

Rhombencephalosynapsis is characterized by fusion of the cerebellar hemispheres with vermician absence. It can be associated with various supratentorial anomalies and rarely with facial and hand anomalies; however, its association with spinal anomalies as well as with cerebellar tonsillar herniations and tectal beaking similar to that seen in Chiari II malformations has not been reported previously. In this article, we report a case with coexistent features of partial rhombencephalosynapsis, Chiari II malformation, and spinal anomalies.

Case Report

Our patient is a 14-year-old boy with normal mental status. He was the first child of a non-consanguineous couple. There was no family history of congenital malformations or any other medical problems. He was a full-term normal vaginal delivery, born with swelling over the upper back. Patient’s mother did not have any antenatal medical supervision with no history of antenatal drug usage. Patient had episodes of cyanotic spells in immediate postnatal period. Patient was operated for cervicothoracic meningomyelocele at the age of 1 year, and now presented with complaints of progressive dorsal kyphoscoliosis and heaviness of right upper and lower limbs, with abnormal gait pattern. On examination, patient’s stature corresponded to that of his father, but having short neck with low-set ears and low posterior hairline, and with uneven shoulder blades and waist, as well as one hip higher than the other (Fig. 1). His neurological examination was unremarkable except having slight right extensor hallucis longus weakness and gait abnormality due to severe kyphoscoliosis, has more limited range of motion while walking with stiffer gait.

On magnetic resonance imaging (MRI) brain, there was mild herniation of cerebellar tonsils in foramen magnum with peg-like appearance (Fig. 2), suggestive of Chiari II malformation, with 4th ventricular compression and proximal dilation of ventricles (Fig. 3). Dysplastic cerebellum showing fusion across midline specially posteriorly suggested rhombencephalosynapsis (Fig. 4), with septal agenesis. Splenium of corpus callosum was hypoplastic. Rest brain imaging was normal.

The MRI spine is suggestive of split cord at C6 to D2 (Fig. 5) with small syrinx in both hemicord, block verte-
brae seen at L2 to L3 levels with duplication of posterior elements and spina bifida. Evidence of split cord is also seen in lumbar region L2 to L3 levels (Fig. 6), cord is low lying and terminates at L4 level with tethering. Scoliosis at cervicothoracic junction and exaggerated dorsal scoliosis and lumbar lordosis is present. Segmentation anomaly in the form of block vertebrae was also noted from C7 to D2 level (Fig. 7). No features suggestive of atlantoaxial dislocation were present. Rest imaging and hematological tests were normal. The patient was advised customized brace and attached to follow-up.

**DISCUSSION**

Rhombencephalosynapsis is a disorder of rhombencephalon. The most characteristic changes associated with the condition include apposition or fusion of the dentate nuclei and cerebellar hemispheric fusion, there is vermian agenesis. Absence of the nodulus results in a key hole-shaped fourth ventricle. Fusion of cerebellar peduncles and colliculi, gray matter heterotopia, stenosis or atresia of aqueduct, hypoplastic or absent inferior olivary nuclei, severe hypoplasia of the cerebellar hemispheres, and an isolated (trapped) fourth ventricle can be associated with the condition.1-10 In our case, cerebellar changes mainly posterior fusion were consistent with partial rhombencephalosynapsis with key hole-shaped fourth ventricle.
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The whole spectrum of supratentorial abnormalities associated with rhombencephalosynapsis is not present, only sepal agenesis, with hypoplastic splenium of corpus callosum being present. Rest abnormalities associated with rhombencephalosynapsis are not seen.

Some case of rhombencephalosynapsis have facial anomalies like hypertelorism, low-set ears, high arched palate,1,11 and hand anomalies.12 In our case, low-set ears and low posterior hairline was present and among supratentorial abnormality dilated ventricle suggestive of arrested hydrocephalus with septal agenesis and dysgenesis of corpus callosum is present. The clinical presentation of rhombencephalosynapsis can range from mental retardation to normal intelligence and is related to the presence and severity associated with supratentorial changes.1-10 The current patient has normal intelligence.

Conversely, Chiari II malformation is a severe and complex anomaly.13-15 It involves rhombencephalon, and spinal column. A meningomyelocele is present in virtually every patient in the lumbar, lumbosacral, or thoracic region. The present case had cervicothoracic meningomyelocele for which he was operated. Other changes associated with Chiari II malformation are small posterior fossa, tectal beaking, stenogyria, spinal segmentation anomalies, and kyphoscoliotic deformity

Rhombencephalosynapsis occurs around gestation period of 41 days, while Chiari II malformation occurs around 28 days.14,16 The embryology of two malformations is thus different and occurs at different gestation periods. Rhombencephalosynapsis represents exaggerated fusion of two cerebellar primordia, while Chiari II is neural tube defect.9,10,13-16 Therefore, such wide spectrum of spinal malformations are not found in patients of rhombencephalosynapsis. Tectal beaking, cerebellar tonsillar herniation, and meningomyelocele are consistent features of Chiari II malformation and are not previously reported with rhombencephalosynapsis; therefore, such coexistence of two embryologically different malformation may represent a new anomaly. In conclusion, we report a patient who presented with features of partial rhombencephalosynapsis with Chiari II features and various spinal anomalies which is such a rare association. It may be a new anomaly of hindbrain with spine.

REFERENCES