Adult rhombencephalosynapsis

Case report

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Rhombencephalosynapsis (RS) is a relatively rare developmental disorder of the cerebellum in which the cerebellar hemispheres are fused across the midline without being separated by a cleft or the vermis. The condition may also be associated with hydrocephalus and other intracranial and extracranial abnormalities. The authors report on the case of a symptomatic adult who was successfully treated with suboccipital decompression and duraplasty. A 39-year-old woman presented with intractable pain radiating from the thoracolumbar column to the occiput. A general examination yielded normal findings and a neurological examination revealed only subtle ataxia of tandem gait. The patient underwent magnetic resonance (MR) imaging, the results of which revealed an absent cerebellar vermis with fusion of the cerebellum and mild hydrocephalus. A cine-MR image obtained to evaluate her cerebrospinal fluid flow (CSF) revealed attenuated flow in the posterior fossa and cerebral aqueduct. Preoperative intracranial pressure (ICP) monitoring demonstrated no elevation of ICP (mean 4.3 mm Hg). The patient consented to undergo suboccipital craniectomy and duraplasty. Despite an increase in postoperative ICP (mean 10.77 mm Hg; difference from preoperative level according to a t-test, p = 0.002), the patient experienced symptomatic relief, which has persisted for 3 years. One year postoperatively, a cine-MR image was obtained, which revealed improvement in the patient’s CSF dynamics. The authors conclude that, although RS may cause altered flow in the adult, their patient has experienced symptomatic relief, suggesting that her pain was related to local pressure in the posterior fossa.

Key Words • cerebellum • developmental anomaly

Abbreviations used in this paper: CSF = cerebrospinal fluid; ICP = intracranial pressure; MR = magnetic resonance; RS = rhombencephalosynapsis.
consistent with RS. The dentate nuclei were apposed and the superior cerebellar peduncles were fused. The MR images revealed a small posterior fossa with downward displacement of the torcula. The fourth ventricle was triangular in shape, the septum pellucidum and the corpus callosum were thinned but preserved, the anterior commissure was present, and there were no abnormalities of the cervical spine. A cine-MR image obtained to evaluate the patient’s CSF flow revealed attenuated flow in the posterior aspect of the foramen magnum and the cerebral aqueduct. A temporary ventriculostomy device was inserted in the patient, who then underwent 24 hours of ICP monitoring, which revealed a mean preoperative pressure of 4.3 mm Hg. Subsequent drainage of CSF at 0 to 1 mm Hg failed to improve her symptoms.

Surgical Procedure and Postoperative Course. The patient underwent a suboccipital craniectomy with autologous duraplasty, which revealed a bulging cerebellum with no vermis. A temporary ventriculostomy device was placed intraoperatively, and revealed a mean postoperative ICP of 10.77 mm Hg, with maximum pressures 5 hours after surgery. This is a significant difference, as measured using a t-test, from the patient’s preoperative ICP (p = 0.002). Despite this rise in ICP, the patient experienced symptomatic relief of her occipital–thoracolumbar pain, which has persisted for 3 years. The patient was able to return to work as a secretary. Results of a neurological examination were unchanged from those of the preoperative examination. A postoperative cine-MR image revealed only minimal improvement in CSF dynamics compared with results of the preoperative imaging study.

Discussion

This case report illustrates most of the following classic pathological findings of RS, which were described by Gross and colleague, absence of the upper cerebellar vermis, and deficiency in the lower vermis with absence of the incisura cerebelli posterior; midline fusion of the cerebellar hemispheres; convergence of the superior and middle cerebellar peduncles; a narrow fourth ventricle; fusion of the superior and medial portions of the dentate nuclei; and fusion of the superior colliculi, with dysplasia or absence of the paleocerebellar nuclei. The advent of MR imaging allowed visualization of several of these pathological findings which were first described in autopsy reports.

Rhomencephalosynapsis results, in part, from abnormal development of the metencephalon, which differentiates into the pons and cerebellum. Normally, the cerebellum develops from symmetrical thickening of the dorsal portions of the alar plates. Neuroblasts in the intermediate zone of the alar plates migrate to the marginal zone and form the cerebellar cortex; other neuroblasts from these plates give rise to the deep nuclei of the cerebellum, the largest of which is the dentate nucleus.

The pathological findings of RS suggest that it is, in part, due to an abnormal commissuration of these neuroblasts. This theory is supported by the association of RS with agenesis of the corpus callosum and hypoplasia of the septum pellucidum which are both commissural disorders. Two genes described by Goodman, et al., comm and robo, are important in the development of midline structures in the central nervous system. Rhomencephalosynapsis may develop from abnormal expressions of these genes during the embryogenesis of the cerebellum. It has also been reported in one case in which there was maternal phencyclidine abuse; however, this relationship is speculative.
Although RS may cause abnormal CSF flow in the adult, our patient experienced relief of her pain after suboccipital decompression without objective evidence of lowered ICP or significantly altered CSF dynamics. The surgical treatment options considered in this case were CSF diversion (ventriculoperitoneal shunt) or posterior fossa decompression. The decision to proceed with decompression was based on the following preoperative data: 1) the cine-MR image documented attenuated CSF flow in the posterior fossa; and 2) the patient’s preoperative ICP was normal and CSF drainage did not provide symptomatic relief.

The increase in the patient’s postoperative ICP was most likely related to pain and was still within the normal range. The patient’s neck pain has been relieved and she has remained shunt free for three years since presentation. This suggests that her symptoms were due to compression of the small posterior fossa rather than a global decrease in ICP.

Fig. 3. Three T2-weighted MR images. Left: Coronal image revealing lateral ventriculomegaly and midline fusion of the dentate nuclei. Center: Axial image demonstrating fusion of the superior cerebellar peduncles. Right: Axial image demonstrating a triangle-shaped fourth ventricle.

References
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