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A Case Report of Rhombencephalosynapsis

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Abstract Rhombencephalosynapsis is a rare cerebellar malformation. It is characterised by dorsal fusion of the cerebellar hemispheres, agenesis or hypogenesis of the vermis, and fusion of the dentate nuclei. A wide variety of associated abnormalities has been reported. To our knowledge, there are less than 50 cases described in the literature so far. However, due to the advent of MRI, this condition has been increasingly reported in recent years. The exact aetiology and associated factors are still not known, therefore, any new case is worth reporting. We describe a case of Rhombencephalosynapsis in a new born and review of the literature.

Key words Cerebellum; Dentate; Rhombencephalosynapsis; Vermis

Case Report

A 19-month-old boy was born at full term, with the birth weight of 3282 g which was at 30th percentile, by caesarian section due to prolonged ruptured of membrane. This baby boy was the third child to a healthy unrelated Spanish parents. The two elder brothers were normal. The mother was healthy all along. There was no history of significant drug intake during the antenatal period. Antenatal ultrasound (US) was done at 16 weeks of gestation but was claimed to be normal.

At birth, head circumference of the child measured 37.8 cm, which was at the 98th percentile, and there was full bulging fontanelles. Therefore, cranial US was performed, revealing dilatation of lateral and third ventricles. Cranial magnetic resonance imaging (MRI) at age 3 days demonstrated agenesis of the cerebellar vermis with fusion of the cerebellar hemispheres. Convergence of the dentate nuclei forming a horseshoe shaped arc across the midline posterior to the forth ventricle could be seen, giving a diamond shape to it (Figure 1a). The lateral and third ventricles were enlarged (Figures 1b & 1c). Forth ventricle was normal in size. Aqueductal stenosis was evident. The corpus callosum was hypoplastic and the tectum was mildly deformed. The septum pellucidum was absent. A ventriculo-peritoneal shunt was inserted. There was reduction in the ventricular size as shown by subsequent follow up computed tomogram (CT) and MRI (Figure 2).

The baby boy had generalised hypotonia. He had no evidence of facial dysmorphism but strabismus. Cranial nerves were essentially normal. The patient had regular follow up and showed some developmental delay. At 19-month-old, he could only walk by holding on with one hand. There was symmetrical crawl. Head control was good. Swallowing was satisfactory. Apart from the strabismus, no visual or hearing problems could be detected. His expressive language was equivalent at 14-month level. Receptive language age was equivalent at 19-month level. No ataxia could be found. No chromosomal abnormality was found.

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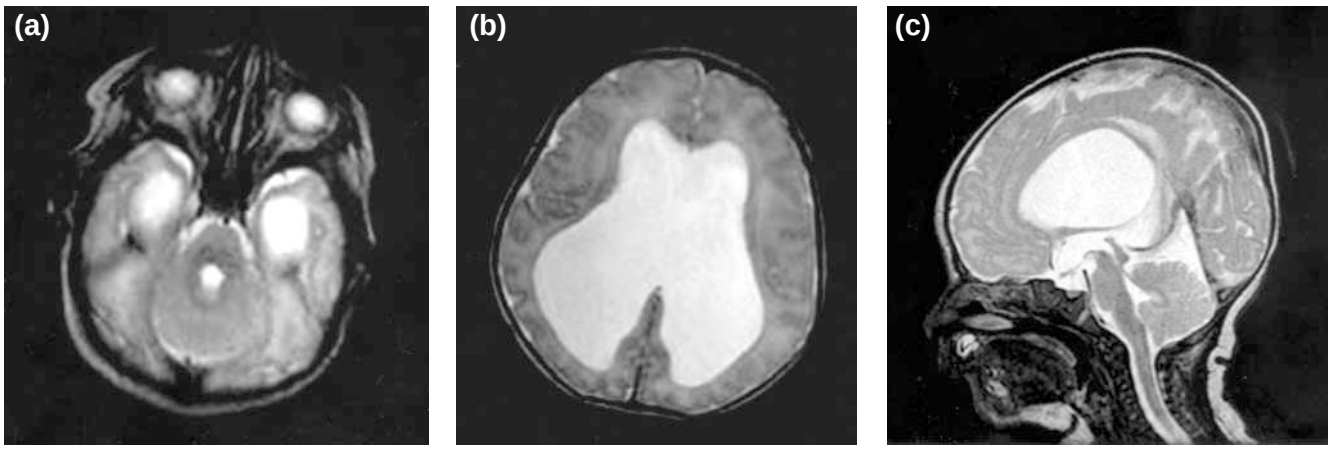

Figure 1. **(a)** Axial MRI T2 weighted images at 3-day-old, showing a diamond-shaped forth ventricle. No intervening vermis was seen between the fused cerebellar hemispheres. The temporal horns were dilated; **(b)** There was hydrocephalus and the septum pellucidum was absent; **(c)** Sagittal MRI T2 weighted image revealed evidence of aqueductal stenosis and hypoplastic corpus callosum.

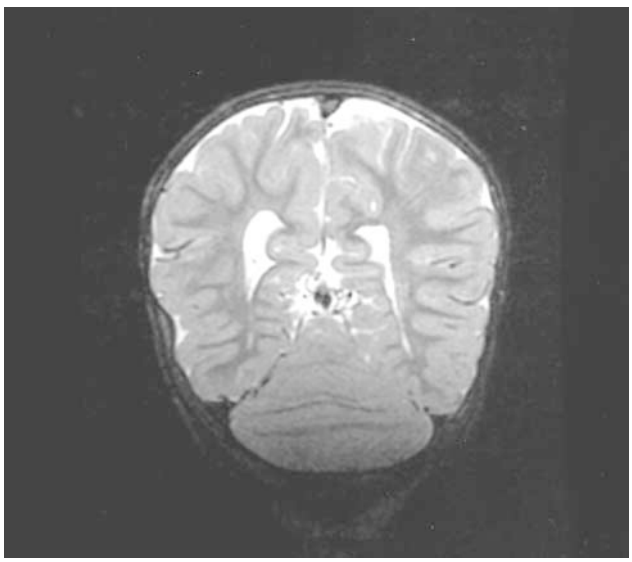

Figure 2 Coronal T2 weighted MRI, taken at 16-month-old, demonstrated the horizontal orientation and continuity of the cerebellar folia across the midline. After the insertion of ventriculo-peritoneal shunt (not shown), there was reduction in the ventriculomegaly.

Discussion

The first case of Rhombencephalosynapsis (RS) was described by Obersteiner[1] in 1914 from an incidental post-mortem examination of a 28-year-old male who died from suicide. The term 'Rhombencephalosynapsis' was later coined by Gross in 1959.[2]

RS is a rare anomaly of the posterior fossa, essentially characterised by vermian agenesis or hypogenesis, fusion of the cerebellar hemispheres and apposition or fusion of the dentate nuclei. There may also be fusion or apposition of the cerebellar peduncles as well as colliculi.[3]

A few reported cases present with an isolated cerebellar malformation.[4] However, various supratentorial midline anomalies are usually associated. The most common reported one is ventriculomegaly.[5] It may be due to the abnormal orientation of the forth ventricle, impairing the cerebrospinal fluid drainage or may be related to aqueductal stenosis, as in our case, which is also a common association with RS.[2,3] Absence of septum pellucidum and fusion of thalami are common; hypoplasia of the commissural system, absence of olfactory tracts and agenesis of the posterior lobe of pituitary gland can be occasionally seen.[6] The corpus callosum can be normal, hypoplastic or dysplastic.[7] Other non-neurological anomalies have also been described.[6]

"Isthmic rhombencephali" is the organising centre for cerebellar development. It is a band of neuroepithelium at the border between the mesencephalon (MES) and the metencephalon (MET).[7] It has been reported that changes like vermian agenesis with dentate fusion, inferior colliculi fusion and aqueductal stenosis may be related to an abnormality in the isthmus.[8] This insult may occur at Carnegie stages 14-18 (28-44 days of gestation)[9] and it is postulated to occur prior to the development of the vermis. This time frame of insult can explain the reported associated midline anomalies.[5] Some authors postulate that the condition can be due to failure of vermian differentiation with undivided cerebellar hemispheres rather than abnormal development of the vermis with subsequent fusion.[5]

Cause is unknown. No teratogenic factor can be identified so far.[3] RS seems to be sporadic in occurrence with no familial recurrence.[3] No particular chromosomal abnormality could be attributed.[3] Romanengo et al suggested the possibility of autosomal recessive inheritance.[10] No metabolic factor was mentioned in previously reported cases though.

Clinical presentation can be variable.[3] Patients usually show early presentation of symptoms. There may be few signs identifying a cerebellar abnormality and sometimes, this can be manifested as global difficulties in motor skills. In fact, the presentation and severity of the clinical disability usually closely correlated to the associated supratentorial anomalies.[11] Most patients die in early life but survival into the third decade has been reported.[9]

The cerebellar abnormalities may be difficult to be detected on computed tomogram (CT) but may be suggested if posterior pointing of the forth ventricle is seen together with large lateral ventricles and absence of septum pellucidum. Sometimes, the forth ventricle may adopt the configuration of a keyhole[3,10] or heart shape.[5]

Magnetic resonance imaging (MRI) is definitely better to delineate abnormality at the posterior fossa and should be the imaging of choice. It allows multiplanar imaging, with superior tissue contrast particularly the gray-white differentiation and absence of beam hardening artifacts at the posterior fossa.[3] It is also better than CT for small key structures such as dentate nuclei, cerebellar peduncles and colliculi which may help identifying the condition.[3] Moreover, the midline abnormalities commonly associated with RS can also be better assessed. In our opinion, apart from the aforementioned advantages, MRI also bears no radiation risk and should be the first imaging modality for investigating patients with non-specific symptoms like developmental delay.

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