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Partial rhombencephalosynapsis and Chiari II malformation

局部後腦窩先天性畸形和II型小腦扁桃體延髓聯合畸形

We report a rare case of partial rhombencephalosynapsis coexistent with Chiari II malformation in a 6-year-old girl and discuss the features of these entities on magnetic resonance imaging.

本文報告一名6歲女童同時患有局部後腦窩先天性畸形以及II型小腦扁桃體延髓聯合畸形的罕見病例，並透過磁共振成像討論病症的特性。

Introduction

Rhombencephalosynapsis (RS) is a rare congenital malformation of the posterior cranial fossa characterised by vermal agenesis or hypogenesis and fusion of the cerebellar hemispheres. About 40 cases have been reported.[1] Partial RS was reported for the first time recently whereby normal development of the anterior vermis and nodulus was noted but part of the posterior vermis was deficient.[2] One case of RS associated with Chiari II malformation has also been reported.[3] To the best of our knowledge, the coexistence of partial RS and Chiari II malformation and their features on magnetic resonance imaging (MRI) have not been reported.

Case report

A 6-year-old girl had spina bifida and hydrocephalus at birth. She was the second child of a non-consanguineous southern Chinese couple. Antenatal examination by a private obstetrician including an ultrasound scan at 22 weeks' gestation was reported to be normal. There was no family history of congenital malformations or any other remarkable medical problems. Her birth at full term was complicated by her large head and the delivery necessitated a Caesarean section. A ruptured myelomeningocele over the lumbosacral region was also noted at birth and there was paucity of lower limb movement. Macrocephaly with features of increased intracranial pressure including dilated scalp veins and sunsetting of the eyes were found. Computed tomographic scan confirmed hydrocephalus, with grossly dilated lateral and third ventricles, and a myelomeningocele at the L5-S1 level. The myelomeningocele was repaired surgically and a ventriculoperitoneal shunt inserted on postnatal day 1 to relieve the hydrocephalus. Chromosomal analysis revealed normal karyotype of 46,XX.

She had recurrent urinary tract infections during infancy. Subsequent ultrasound study of the urinary tract revealed a single horseshoe kidney with bilateral hydronephrosis and a mildly irregular renal contour. Further dimercaptosuccinic acid study revealed scarring of the left side of the single kidney. Long-term low-dose prophylactic antibiotic therapy was prescribed.

In the ensuing years, she was found to have global developmental

Key words:
Arnold-Chiari malformation;
Cerebellum;
Child;
Magnetic resonance imaging;
Rhombencephalon

關鍵詞：
小腦扁桃體延髓聯合畸形；
小腦；
兒童；
磁共振成像；
菱腦

Hong Kong Med J 2005;11:299-302

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delay, cerebellar ataxia, and mild dystonia. She entered a special child care centre at the age of 3 years, where she received intensive training. There was a steady improvement and satisfactory progress in her development.

Magnetic resonance imaging at 5 years of age revealed absence of the inferior cerebellar vermis (Fig 1a), hypoplasia of the superior cerebellar vermis, and partial fusion of the two cerebellar hemispheres (Fig 1b). The nodulus was present, although hypoplastic (Fig 1b). There was partial fusion of the thalami (Fig 2a), but the dentate nuclei, superior and inferior colliculi were not fused. The folia were transversely oriented and extended across the whole of the cerebellum (Fig 1). There was a towering appearance of the fused cerebellum at the region of the tentorial hiatus. In addition, the lateral aspects of the cerebellum were displaced anterolaterally, 'wrapping' around the brainstem, which was hypoplastic (Fig 1a). The fourth ventricle was slightly dilated and had a keyhole shape. The tegmental plate appeared beak-shaped (Fig 2b). The posterior cranial fossa was small and the cerebellar tonsils extended below the level of the foramen magna into the spinal canal (Fig 2b). Supratentorially, the brain was grossly abnormal. The ventricles were decompressed such that the ventricular walls were nearly coapted. The corpus callosum was markedly hypoplastic and distorted. The septum pellucidum was not apparent. There was also marked reduction of cortical white matter. Gyral pattern in the parieto-occipital region was abnormal and the appearance was in keeping with stenogyria. In addition, there was interdigitation of gyri across the interhemispheric fissure. The medial temporal lobe and the hippocampi appeared hypoplastic. These features were consistent with the diagnosis of partial RS with a Chiari II malformation.

Discussion

Rhombencephalosynapsis was first described by Obersteiner in 1914[4] and the first case diagnosed by MRI was reported in 1991.[5] It has been estimated that RS occurs with a frequency of 0.13% in the paediatric population[6] but the true frequency could be higher than the estimate, due to an increasing awareness of the malformation, as well as advancements in magnetic resonance technology and its increasing availability. Both the new entity of partial RS and coexistent RS and Chiari II malformation were described for the first time within the last 2 years.[2,3] Our findings of the presence of the anterior vermis and nodulus, absence of the posterior vermis, and partial fusion of the cerebellar hemispheres and thalami are similar to that of the case reported recently by Demaerel et al.[2] In

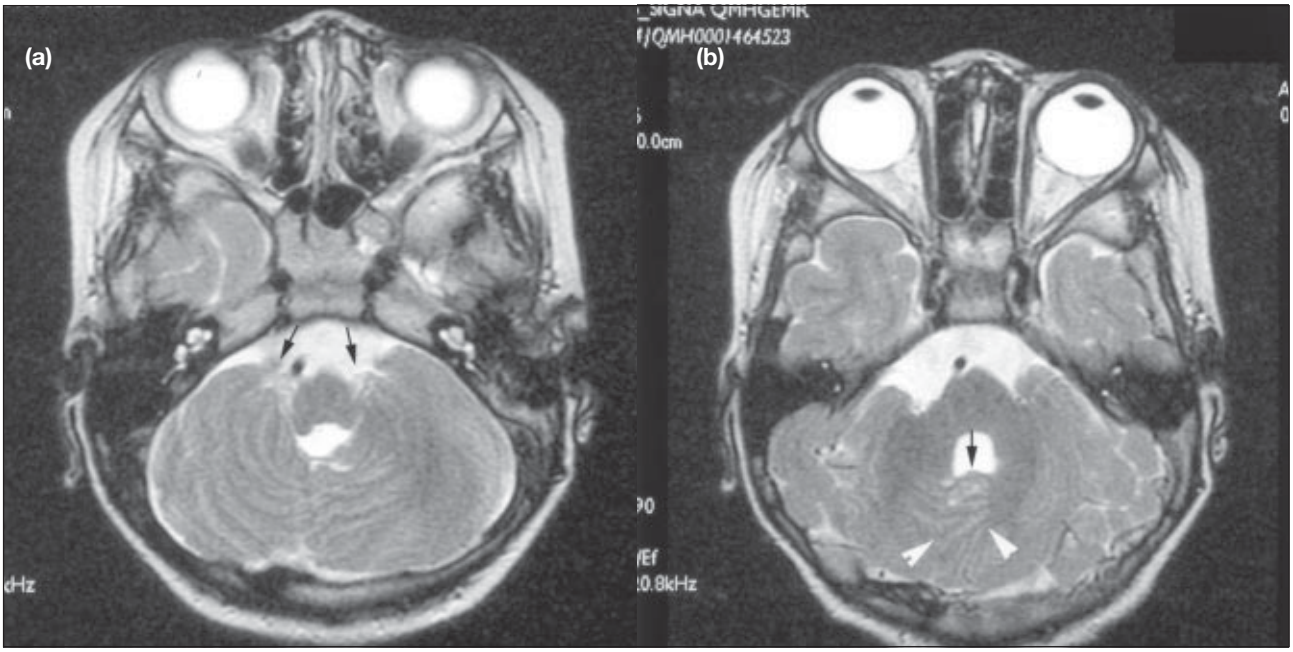

Fig 1. (a) Axial T2-weighted magnetic resonance image showing absence of the inferior cerebellar vermis, transversely oriented cerebellar folia, and 'wrapping' of the cerebellum around the brainstem (arrows); (b) axial T2-weighted magnetic resonance image showing hypoplastic superior cerebellar vermis and nodulus (arrow) and fusion of the cerebellar hemispheres (arrowheads)

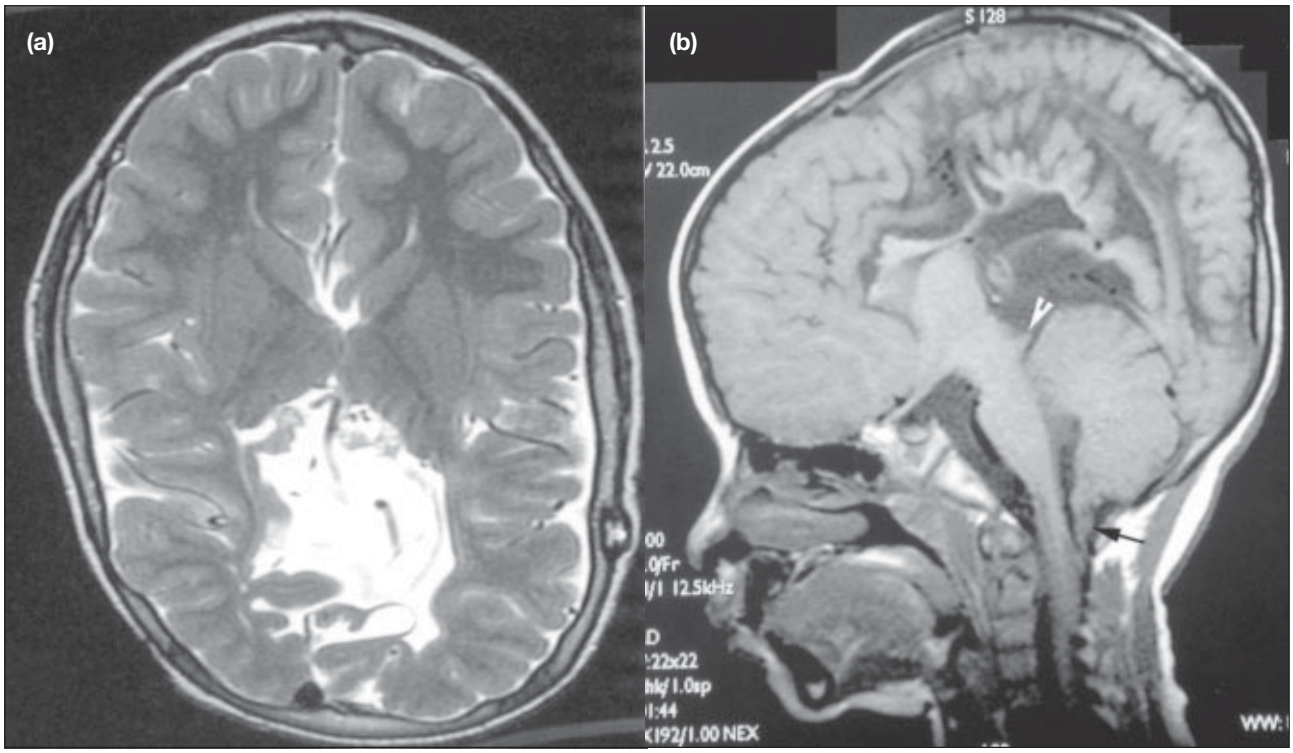

Fig 2. (a) Axial T2-weighted magnetic resonance image showing partial fusion of the thalami; (b) sagittal T1-weighted magnetic resonance image showing features of the Chiari II malformation; a small posterior fossa with herniation of the cerebellar tonsils through the foramen magnum (arrow) and beaking of the tectum (arrowhead)

contradistinction, the tonsils were present and herniated through the foramen magnum in our patient and there was no fusion of the colliculi. The recent view of cerebellar development considers the cerebellar premordium an unpaired structure with the development of the posterior vermis followed by the anterior vermis.[7,8] Rhombencephalosynapsis is therefore attributed to a failure of vermian differentiation rather than an abnormal fusion of the two cerebellar hemispheres.[8] The theory that the posterior vermis develops before the anterior vermis does not support the findings of absent posterior vermis in the presence of an anterior vermis in both cases of partial RS.[2] It is possible that the defect seen in partial RS is due to some intrinsic inability of the posterior vermis to respond to inductive signals for its differentiation.

The theories for the pathogenesis of RS include genetic defects; it has been suggested that RS probably represents an underexpression of a dorsalising organiser gene.[9] Cerebellar midline and caudal midbrain abnormalities resembling that seen in RS have been identified in a mutant mouse model Dreher (dr) that has a homozygous mutation in the *Lmx1a* gene.[10] Further characterisation and study of such dorsalising factors may shed light on the pathogenesis of RS.[11]

It has been suggested that RS is sometimes misinterpreted as Chiari II malformation by the unwary because of the rarity of RS and their overlapping imaging features: ventriculomegaly, hypoplasia of the corpus callosum, and a relatively small-sized posterior fossa leading to an impression of downward pointing of the cerebellar tonsils and a small fourth ventricle.[6] In our patient, as in the previously reported case of coexisting RS and Chiari II malformation,[3] there was a myelomeningocele that is almost invariably present in Chiari II malformations. Other associated features of Chiari II malformation, such as beaking of the tectum, gyral interdigitation through a fenestrated falx, and displacement of the cerebellum to 'wrap' round the brainstem, were also seen in our patient. In addition, an 18-fold increase in the incidence of fused kidneys with meningomyelocele was reported[12] and this coincides with the occurrence of a horseshoe kidney in our patient. The reason for the association is not entirely clear. It has been speculated that the deformity of the spine in meningomyelocele may bring the developing kidney into closer proximity than normal because they migrate caudally to assume their adult position, encouraging fusion to occur.[12]

This case report highlights the superiority of MRI

in diagnosing posterior fossa malformations, especially in the presence of complex pathology.

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