

Case 8962

Partial rhombencephalosynapsis

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Section: Neuroradiology

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Patient: 4 year(s), male

Clinical History

A 4-year-old boy, born prematurely at 29 weeks (twin pregnancy), with periventricular leukomalacia and epilepsy underwent brain MRI. Neurological examination showed severe developmental retardation with axial hypotonia, spastic tetraparesis and convergent strabismus.

Imaging Findings

Cranial MRI revealed typical aspects of partial rhombencephalosynapsis with vermian hypoplasia, midline fusion of the cerebellar hemispheres and transversely oriented folia and fissures (Fig.1). There was also mild dilatation and dysmorphism of the ventricular system. The septum pellucidum was absent. The hippocampi were malrotated and had vertical orientation (Fig. 2) Associated periventricular cystic leukomalacia.

Discussion

Rhombencephalosynapsis (RS) is a rare congenital defect of the cerebellum classically characterised by vermian agenesis or hypogenesis, fusion of the hemispheres, and closely apposed or fused dentate nuclei [1]. It is now considered to result from an absence of division of the cerebellar hemispheres, following an insult between the 28th and 44th day of gestation (i.e., before the formation of the vermis) [2].

Other features have also been described such as fusion of the thalami and cerebral peduncles, malrotated hippocampi, corpus callosum agenesis, hypoplastic chiasm, absence of the septum pellucidum, ventriculomegaly, agenesis of the posterior lobe of the pituitary and cortical malformations. Musculoskeletal, cardiovascular, urinary tract, and respiratory abnormalities have

been reported [2, 3].

Typical symptoms consist of swallowing difficulties, delayed motor acquisitions, muscular hypotonia, spastic quadriparesis, cerebellar signs including dysarthria, gait ataxia, abnormal eye movements, and seizures and hydrocephalus [3].

The major MRI signs consist of fused cerebellar hemispheres, with absent or hypoplastic vermis, narrow diamond-shaped fourth ventricle and fused dentate nuclei. In a minority of cases, partial RS has been identified by MRI [3], demonstrating the presence of the nodulus and the anterior vermis and absence of part of the posterior vermis with only partial fusion of the hemispheres in the inferior part [3].

Other cerebellar malformations involving vermian agenesis or hypoplasia include the Dandy-Walker continuum, Joubert syndrome, tectocerebellar dysraphy or pontocerebellar hypoplasias, and are now easily distinguished from RS by both brain MRI and morphology [1, 2].

Final Diagnosis

Partial rhombencephalosynapsis

Differential Diagnosis List

Dandy-Walker continuum, Joubert syndrome, Tectocerebellar dysraphy or pontocerebellar hypoplasias

Figures

Figure 1 Figure 1

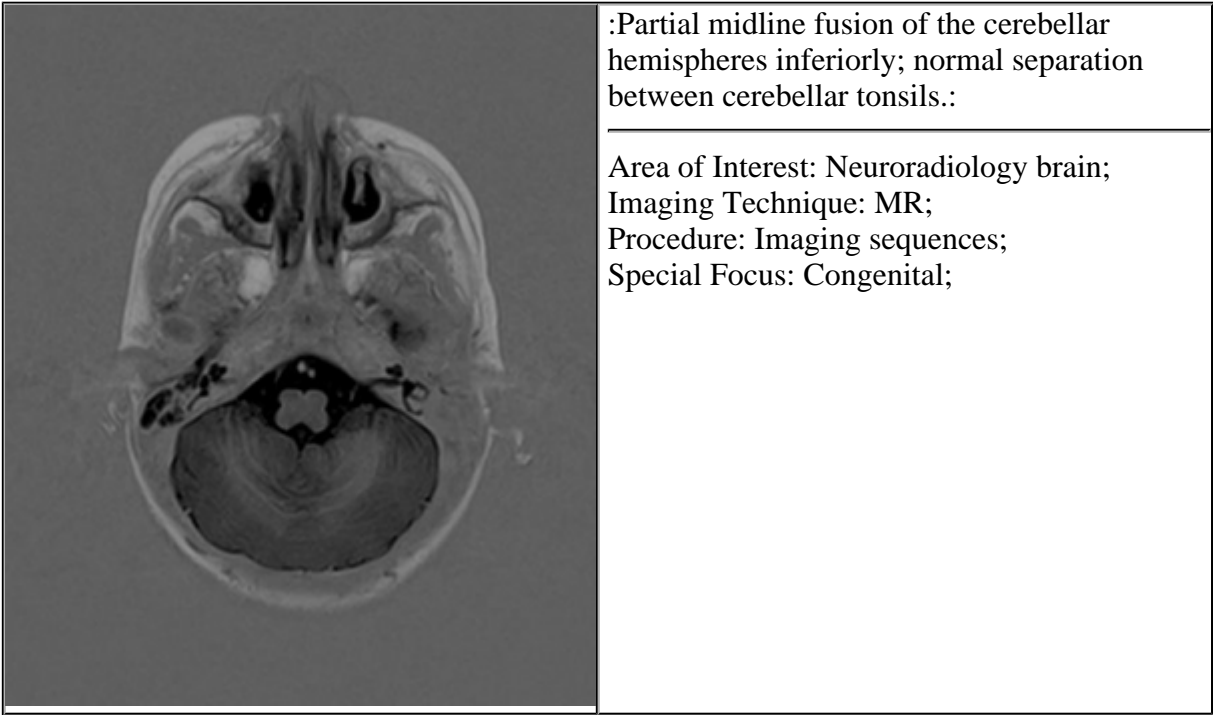
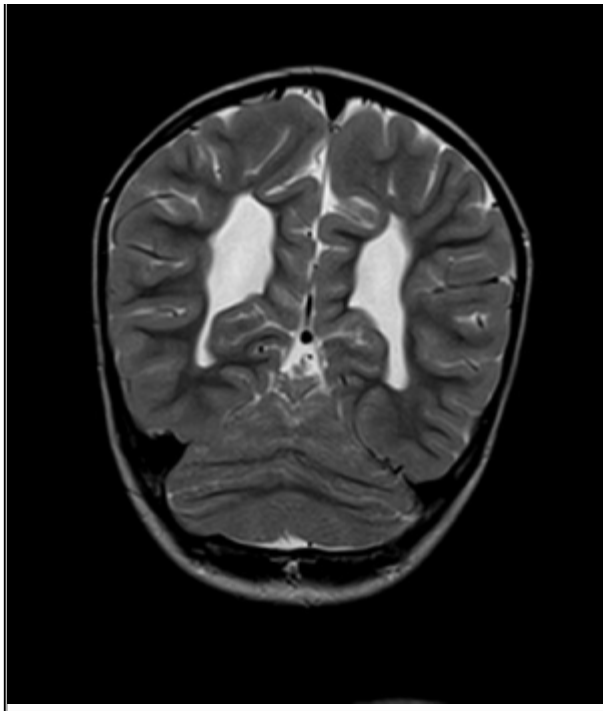


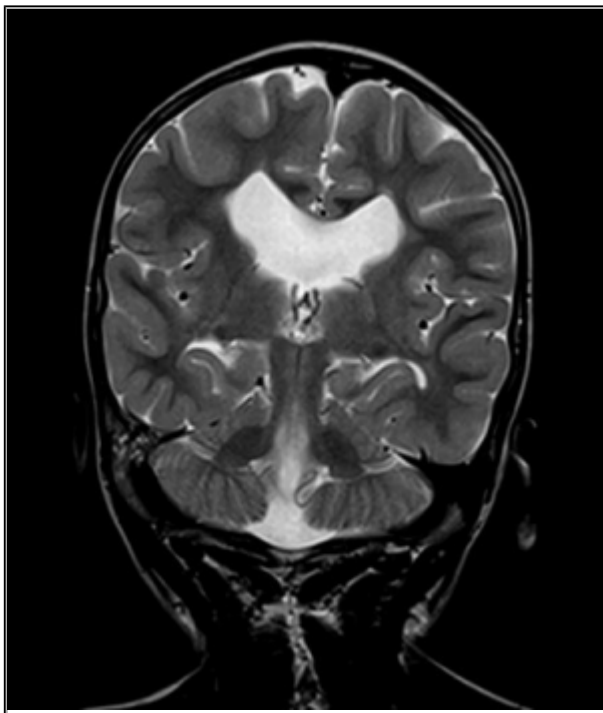
Figure 2 Figure 2



:Transversely oriented folia; dilatation and dysmorphism of the ventricular system.:

Area of Interest: Neuroradiology brain;
Imaging Technique: MR;
Procedure: Imaging sequences;
Special Focus: Congenital;

Figure 3 Figure 3



:Malrotated an vertically orientated hippocampi.:

Area of Interest: Neuroradiology brain;
Imaging Technique: MR;
Procedure: Imaging sequences;
Special Focus: Congenital;

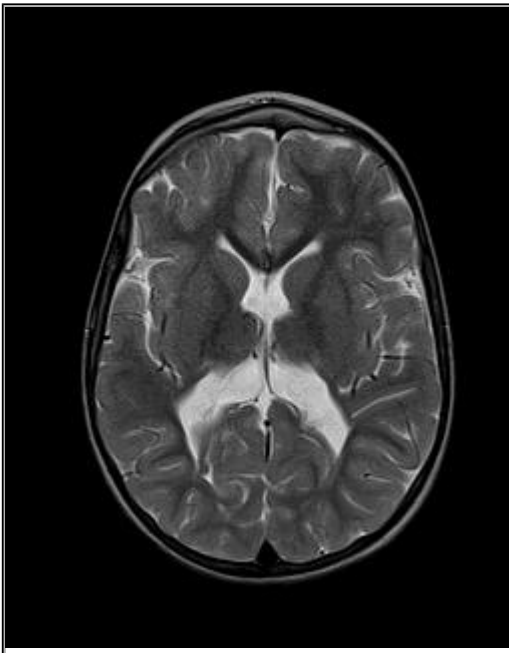
Figure 4 Figure 4



:Cerebellar tissue is present in the midline, but it does not have the normal vermian lobular configuration;:



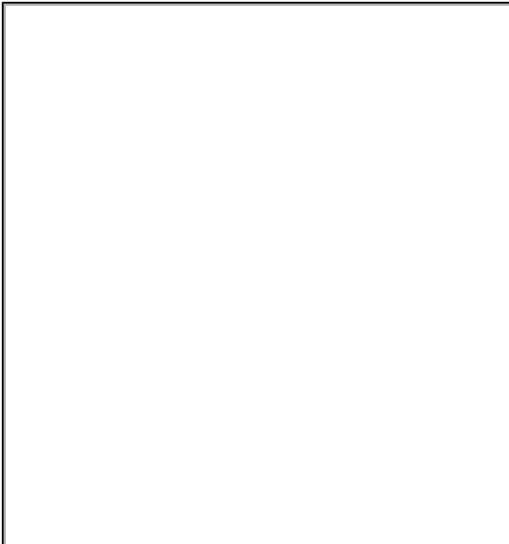
Figure 5



:Ventriculomegaly with dilatation of the trigones and an irregular ventricular outline;;

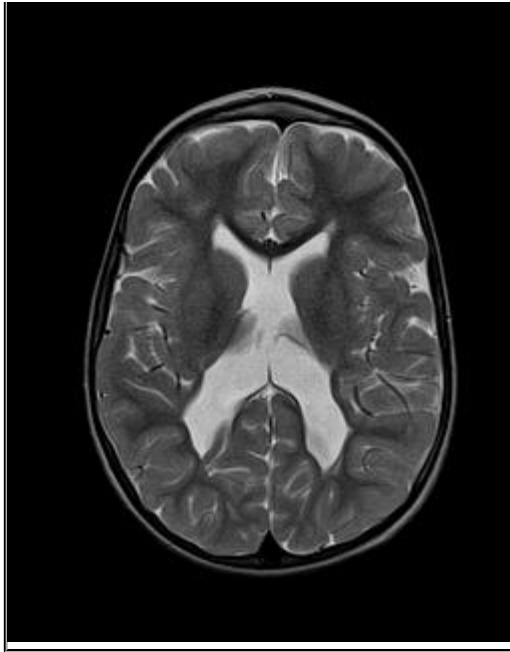
Area of Interest: Neuroradiology brain;
Imaging Technique: MR;
Procedure: Imaging sequences;

Figure 6



:Ventriculomegaly with an irregular ventricular outline;;

Area of Interest: Neuroradiology brain;
Imaging Technique: MR;
Procedure: Imaging sequences;



MeSH

Cerebellum [A08.186.211.212]

Part of the metencephalon that lies in the posterior cranial fossa behind the brain stem. It is concerned with the coordination of movement.

Rhombencephalon [A08.186.211.132.810]

That part of the brain stem constituting the MEDULLA OBLONGATA (myelencephalon) and PONS (metencephalon).

Infant, Newborn, Diseases [C16.614]

Diseases of newborn infants present at birth (congenital) or developing within the first month of birth. It does not include hereditary diseases not manifesting at birth or within the first 30 days of life nor does it include inborn errors of metabolism. Both HEREDITARY DISEASES and METABOLISM, INBORN ERRORS are available as general concepts.

Cerebellar Diseases [C10.228.140.252]

Diseases that affect the structure or function of the cerebellum. Cardinal manifestations of cerebellar dysfunction include dysmetria, GAIT ATAXIA, and MUSCLE HYPOTONIA.

Nervous System Malformations [C10.500]

Structural abnormalities of the central or peripheral nervous system resulting primarily from defects of embryogenesis.

References

- [1] Anthony T. Yachnis (2002) Rhombencephalosynapsis with massive hydrocephalus: case report and pathogenetic considerations Acta Neuropathologica 103 :301-304
- [2] Philippe Demaerel, Catherine Morel, Lieven Lagae, and Guido Wilms (2004) Partial

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[3] Pasquier L, Marcorelles P, Loget P, Pelluard F, Carles D, Perez MJ, Bendavid C, de La Rochebrochard C, Ferry M, David V, Odent S, Laquerrière A (2008) Rhombencephalosynapsis and related anomalies: a neuropathological study of 40 fetal cases Acta Neuropathologica 117: 185-200

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