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Partial rhombencephalosynapsis: prenatal MR imaging diagnosis and postnatal follow up

Katarina M. Koprivsek[1], Aleksandra S. Novakov-Mikic[2], Milos A. Lucic[1], Dusko B. Kozic[1], Viktor E. Till[3] and Zoran Belopavlovic[4]

[1]Diagnostic Imaging Centre, Institute of Oncology of Vojvodina, Sremska Kamenica, Serbia; [2]Department of Gynecology and Obstetrics, [3]Centre of Radiology, [4]Department of Gynecology and Obstetrics, Clinical Centre of Vojvodina, Novi Sad, Serbia

Abstract

We present a case of partial rhombencephalosynapsis, diagnosed by magnetic resonance imaging (MRI), in fetus aged 27 gestational weeks, in a dizygotic twin pregnancy. The distinctive MRI features of this cerebellar malformation (segmental hypogenesis of the cerebellar vermis, partial fusion of the cerebellar hemispheres and dentate nuclei) without associated cerebral abnormalities were confirmed by 32-weeks prenatal and 3-months postnatal MRI studies. At the age of 12 months the affected twin had a slight delay in psychomotor development, mild hypotonia with normal cognitive development. To the authors' best knowledge, this is the first report of a fetal case with isolated partial rhombencephalosynapsis. Its MRI features enlarges the narrow spectrum of uncommon variants of rhombencephalosynapsis, and allow an accurate differentiation from other vermian and cerebellar anomalies with less favorable postnatal outcome.

Key words: Cerebellar malformation; rhombencephalosynapsis; prenatal diagnosis; magnetic resonance imaging.

Introduction

Rhombencephalosynapsis (RS) is a sporadic cerebellar malformation consisting of vermian agenesis or severe hypogenesis, fusion of the cerebellar hemispheres and apposition or fusion of the dentate nuclei (1). Since the first Obersteiner's report in 1914, not more than 50 cases of the classical form of the RS have been described; with approximately 36 cases diagnosed by magnetic resonance imaging (MRI). The unique case of partial RS was described by Demaerel and colleagues, as partial fusion of the inferior parts of cerebellar hemispheres and dentate nuclei. The cerebellar malformation of their young patient was in conjunction with brain and extracranial anomalies and severe neurological and cognitive impairment (2). To the best of authors' knowledge, prenatal diagnosis of isolated partial RS with favorable postnatal outcome, has never been reported.

Case report

A 29-year-old primigravida, with no relevant medical history, was referred to MRimaging unit, at 27 weeks of dizygotic twin pregnancy, after routine ultrasound (US) examination, performed at 21th gestational week suggested single-lobed cerebellum in the second twin. Fetal MRI was performed on a 1.5T-unit. Four-millimeter-thick half Fourier single-shot fast-spin-echo (760/104/1;TR/TE/excitations) T2-weighted images were obtained in three reference planes.

The fetal MRI study in the second twin revealed mild ventriculomegaly and corpus callosum thinning. The posterior fossa and fourth ventricle were of the normal size, with the rounded fastigial point. Cerebellar diameters were smaller then expected for gestational age. There was partial fusion of the inferior part of the cerebellar hemispheres and dentate nuclei. The anterior vermis appeared normally developed, but posterior vermis was deficient. The primary and secondary fissures could be recognized. Uvula, pyramis and prepyramidal fissure couldn't be identified (Fig. 1A and B). In the first twin, fetal MRI confirmed normal brain finding. Due to possible development of the "late-appearing" cerebral malformation in the second twin, it was decided to perform MRI-follow up.

The second prenatal MRI study (performed at 32 weeks' gestation) revealed normalization of the lateral ventricles volume and significant cerebellar

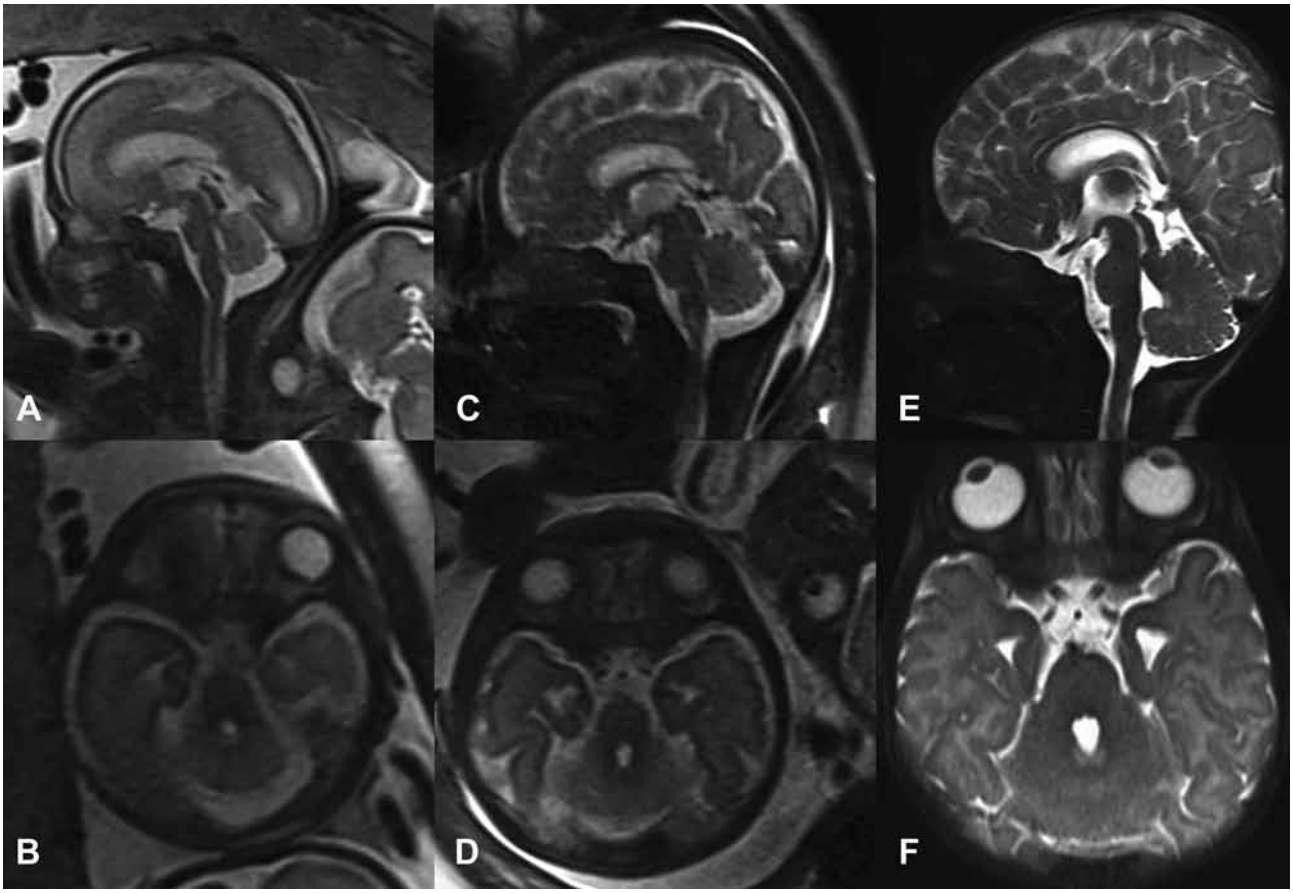

FIG. 1. — Partial rhombencephalosynapsis. Prenatal and postnatal MR images. A and B. Sagittal (A) and axial (B) MRI T2W images obtained at 27 weeks of gestation show normal sized posterior fossa and fourth ventricle, with rounded fastigial point. The cerebellum is smaller than expected for this gestational age, with partially fused inferior portions of the hemispheres and dentate nuclei. Anterior vermis appears normally developed, while posterior vermis is deficient. In the sagittal plane, the primary fissure can be recognized; the secondary fissure is present, but tiny; C and D. Follow-up sagittal (C) and axial (D) MRI T2W images obtained at 32 weeks of gestation show significant cerebellar growth. Partial fusion of the cerebellar hemispheres and abnormal vermian folial pattern are more prominent. The vermian fissures are more pronounced, but still shallow, compared to the depth of the vermian fissure in the healthy, first twin (not shown); E and F. Postnatal, sagittal (E) and axial (F) T2-weighted images obtained at 3 months of age confirm the prenatally detected cerebellar and vermian pathology. Except for the slightly thinner corpus callosum, the remainder of the supratentorial brain exhibited none of the abnormalities, commonly found in association with the classical form of rhombencephalosynapsis.

growth. Previously detected cerebellar pathology and abnormal vermian folial pattern become more obvious (Fig. 1C and D). The vermian fissures were slightly more pronounced, but still shallow comparing to the fissures depth of the healthy, first twin.

At 39 weeks' gestation, male neonates were born by Caesarian section. Body biometric measurements for both twins were within normal limits. The karyotypes were normal, as well as neurological status and findings in the first twin. In the second twin, neonatal neurological examination revealed only mild hypotonia. Body anomalies were not detected. At 3 months of age, control MRI was performed, using the same system of the prenatal studies. The study confirmed the prenatal diagnosis of partial rhombencephalosynapsis without associated cerebral malformations (Fig. 1E and F).

Clinical findings at 6 months and 1 year were age-appropriate, except of mild hypotonia and slight delay in motor development. No seizure episodes were reported and the electroencephalographic pattern was normal. Battelle's developmental inventory test did not identify any delay in cognitive development.

Discussion

Rhombencephalosynapsis (RS) is a sporadic cerebellar malformation characterized by agenesis of the vermis, fusion of the cerebellar hemispheres, dentate

nuclei and superior cerebellar peduncles leading to the characteristic keyhole-shaped appearance of the fourth ventricle. In majority of cases cerebellar anomalies are associated with wide range of cerebral malformations (dysgenesis of corpus callosum and anterior commissurae, fusion of the inferior colliculi, cerebral peduncles or thalami and hydrocephalus) (1-3). Most of the reported cases had uniform imaging features of the cerebellum, vermis and dentate nuclei; they differed only in number and type of associated cerebral and extracranial anomalies (3). Nevertheless, the uniformity of the cerebellar morphology in RS have been questioned, after Demaerel and colleagues described their unique case of partial RS (2).

The prenatal diagnosis of RS is very important for prenatal counseling, as this malformation is uncorrectable and in majority of cases carries a poor prognosis. The postnatal clinical presentation is variable, and includes truncal and/or limb ataxia, hypotonia, abnormal eye movements, epilepsy, developmental delay and psychiatric disorders. Long-term cognitive outcome in RS is unpredictable, but cognitive functions are mostly impaired (4). According to Toelle's group, additional CNS-anomalies and hydrocephalus could influence the long-term cognitive outcome (1).

To date, only three studies have evaluated prenatal imaging in fetuses with classical form of RS. They reported US/MRI findings in eight fetal cases with classical RS, associated with cerebral and extracranial anomalies. In all cases, postnatal outcome was poor; including death in the neonatal period or early infancy, in more than a half of the group (5). On the contrary, our case is a prenatally diagnosed variant of the RS with distinctive MRI features (segmental vermian hypogenesis, partial fusion of the cerebellar hemispheres and dentate nuclei without associated cerebral malformations) compatible with a favorable outcome.

Presented case contributes to the enlargement of the narrow spectrum of uncommon variants of RS. The recognition of presented distinctive prenatal-MRI features allows a differentiation from the other vermian/cerebellar anomalies with less favorable outcome.

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Katarina Koprivsek, M.D., Ph.D.,
Diagnostic Imaging Centre,
Institute of Oncology of Vojvodina,
Institutski put 4,
21204 Sremska Kamenica (Serbia).
E-mail: katarina.koprivsek@gmail.com