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 extra-
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 hydro-
cephalus) (1-3). Most of the reported cases had uni-
form imaging features of the cerebellum, vermis and
dentate nuclei; they differed only in number and type
of associated cerebral and extracranial anom-
alties (3). Nevertheless, the uniformity of the cere-
bellar 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 uncor-
rectable and in majority of cases carries a poor prog-
nosis. 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 func-
tions 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 extracra-
nial 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.

Acknowledgments

We thank the radiologists Milan Bundalo, Zeljko Babic
and Bojan Grujicic, of the Diagnostic Imaging Center in
Sremska Kamenica for technical assistance in data
acquisition and their excellent support. We also express
our sincere gratitude to our little patient and his family
who participated in the study.

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