Original Investigation

Long Term Outcomes of Fetal Posterior Fossa Abnormalities Diagnosed with Fetal MRI

Şeker et al. Fetal Posterior Fossa Anomalies

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Abstract
Objective: The diagnosis of posterior fossa abnormalities (PFA) in the intrauterine period; and pregnancy outcomes are still controversial. PFA is generally referred to as maternal-fetal medicine specialists. The primary purpose of PFA diagnosis is to screen for other accompanying abnormalities, provide prognostic information to families, and discuss the termination option.
Material and Methods: This retrospective study was conducted with patients diagnosed with PFA between January 2013 and September 2020 in the tertiary Perinatology Clinic. All patients were routinely performed second-trimester ultrasound screening, and the definitive diagnosis was made by fetal MRI in the presence of a suspected anomaly.
Results: Between 2013 and 2020, 164 fetal MRIs were performed for fetal abnormalities, and 22 fetuses were diagnosed with a PFA on fetal MRI. Considering the fetal MRI indications of the cases, four patients (18%) were diagnosed with Mega Cisterna Magna (MSM), Two patients were diagnosed with rhomboencephalosynapsis, thirteen patients were diagnosed with Vermian Hypoplasia-Dandy-Walker variant. In the remaining two patients, those with neural tube defects and lumbosacral NTD are still alive. However, iniencephaly was detected in the other patient and died in the postnatal period.
Conclusion: Diagnosis of PFA abnormalities is complex, and it is a condition that cannot be predicted clearly which patients will have a good prognosis and which will have a bad prognosis. The prognosis is not affected by maternal and fetal factors and allows the recognition of additional accompanying abnormalities. Fetal MRI is an imaging method that can provide retrospective examination and research, especially in pregnancies with poor prognoses.
Keywords: Antenatal ultrasound, Dandy-Walker, fetal anomaly, Fetal MRI, obstetric ultrasound, posterior fossa abnormalities

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DOI: 10.4274/jtgga.galenos.2022.2022-7-13

Received: 20 August, 2022 **Accepted:** 09 November, 2022

Introduction

The cerebellum is the center for motor coordination, execution, and higher cognitive functions (1, 2). In addition, the cerebellum may be associated with impaired spatial navigation and musical learning ability, as well as acting as a modulator for the motor, sensory, cognitive, emotional, and autonomic areas (1, 3). Therefore, the effect of this anomaly on child development should be known.
The diagnosis of posterior fossa abnormalities (PFA) in the intrauterine period; and pregnancy outcomes are still controversial. PFA is generally referred to as maternal-fetal medicine specialists. The incidence of PFA is approximately 1:5000 in live births (4). PFA is generally divided into two groups: primer and seconder Primary fossa abnormalities are divided into two groups: Dandy-Walker Continuum (DWC) and Other Abnormalities of the Cerebellar Hemispheres (5).

The cerebellum has specific stages of development, so its development is different from other brain organs (6). Basic ultra-sonographic (USG) evaluation of PFA is usually performed in the second trimester via transabdominal from a transverse view of the fetal head at the level of the trans cerebellar axial plane; typically, measurements of the anteroposterior diameters of the cisterna magna are used for the evaluation of the posterior fossa (7). Recently, the mid-sagittal cranial plane has been highlighted as an essential diagnostic step in the antenatal imaging of PFA, emphasizing that the best assessment of PFA can be achieved by analysis of a combination of mid-sagittal and axial sections (7).

Abnormal PF size is an essential sign in the diagnostic triage of fetuses with central nervous system malformations. It is enlarged in DWC and narrowed in Chiari II malformation (CM-II). The sagittal expansion of the PF is usually associated with the elevated position of the tentorium and its occipital insertion into the torcular herophili region. In PFA, fetal magnetic resonance imaging (MRI) can be used when the diagnosis cannot be made by USG examination or to confirm the diagnosis. It has been emphasized that diagnoses can change by 70% after fetal MRI (8).

There are limited publications in the literature on long-term postnatal outcomes of patients diagnosed with PFA by fetal MRI. The primary purpose of this article is to analyze the neurological outcomes of children diagnosed with PFO prenatally.

Material and Methods

This retrospective study was conducted with patients diagnosed with PFA between January 2013 and September 2020 at the Perinatology Clinic of Ankara University School of Medicine, Department of Obstetrics and Gynecology. The Institutional Review Board approved the study. All patients were routinely performed second-trimester ultrasound screening, and the definitive diagnosis was made by fetal MRI (Philips Ingenia 1.5 Tesla) in the presence of a suspected anomaly. In addition, referral patients diagnosed with any anomaly in another center were also included in the study. Ultrasonographic examination of all patients was performed with Voluson 8 GE Healthcare Ultrasound, Milwaukee, WI, USA), 4-8 Mhz convex transabdominal transducer, and 5-9 Mhz transvaginal transducer. Demographic data of patients diagnosed with PFA were analyzed from hospital records, and an experienced perinatologist reviewed ultrasonographic images. The images were compared with fetal MRI recordings. Amniocentesis was performed to mothers who accepted amniocentesis. When an abnormality was detected, information about the prognosis was given by the perinatology specialist. The termination of pregnancy (TOP) was applied to the parents who chose to terminate the pregnancy. In our country, there is no upper limit of gestational week for termination of pregnancy in the presence of severe abnormalities. Follow-up examinations were performed biweekly for up to 38 weeks. Some patients who did not undergo termination underwent MRI or computed tomography (CT) after birth to confirm the diagnosis. US results obtained at the time of diagnosis and accompanying abnormalities encountered during follow-up were recorded. Delivery type and week, postnatal neurologic development results of the fetuses were recorded. Detailed neurological and developmental examination could not be performed because the living babies were residing in different provinces. Information about the vision, speech and motor functions of the patients was obtained from the parents by telephone. It was learned whether he was able to use a mechanical ventilator or whether he could meet his own needs. This study was conducted after the Ankara University Cebeci Hospital Clinical Research Ethics Committee's gave approval and written informed consent was obtained from all participants. The records of patients who underwent postnatal MRI examination were also reviewed.

Statistical Analysis

We used the IBM SPSS 26.0 for Windows (SPSS Inc., Chicago, IL, USA) statistical package for statistical evaluation of our research data. A descriptive analysis of the records of the patients was made and a table was created. Categorical variables were calculated as frequency and percentage. Continuous perinatal data were summarized using the median, mean, and SD, and categoric factors were summarized using proportions.

Results

Between 2013 and 2020, 164 fetal MRIs were performed for fetal abnormalities, and 22 fetuses were diagnosed with a PFA on fetal MRI. The mean age of the patients was 23.68 (21-43). The mean gestation age was 26.34(19-36) weeks during MR imaging. In the termination group, the mean gestational week was 26.27 (23-27). In the non-termination group, the mean gestational age at birth was 36.97 (35-40) weeks, and the mean birth weight was 2911.76 (425-4100) grams. The ages of the participants differed from 1 to 8. The demographics of the patients are given in Table-1.

Considering the fetal MRI indications of the cases, four patients (18%) were diagnosed with Mega Cisterna Magna (MSM), and the neurological examination of all patients was within normal limits. Two patients were diagnosed with rhomboencephalosynapsis, the first patient had preterm labor, and subsequently, the fetus died at 24 weeks, and the second patient had additional esophageal atresia, and her/his neurological examination is now within normal limits. Fourteen patients were diagnosed with DWC, and four of them were terminated. One pregnant woman presented

with abruptio placentae and ultrasonography revealed that the fetus had no cardiac activity. Two of our patients died in the postnatal period, and both had additional abnormalities. One had a double out right ventricle (DORV), and the other had interrupted aorta and diastematomyelia. There were six surviving infants; two of them had additional abnormalities besides neurological abnormalities. One of them had cerebellar hypoplasia, and the baby had a speech delay and balance problems. The other has ventriculomegaly (31 mm) detected in the intrauterine period and currently has cerebral palsy. Neurological examination of four patients with isolated PFA was within normal limits. In the remaining two patients, those with neural tube defects and lumbosacral NTD are still alive. However, iniencephaly was detected in the other patient and died in the postnatal period. Abnormalities of fetus and results of whose long term is shown Table-2.

Discussion

Fetal MRI is generally preferred in patients whose definitive diagnosis cannot be made after second-trimester sonography, and it gives undetected findings with ultrasound at a rate of 60% (8). In our case, fetal MRI was performed between 26 and 34 weeks of gestation, in line with the literature.

One of the main results of our study is that if fetal PFA is isolated, the neurologic examination is usually within normal limits. If there is a fetal PFA, additional abnormalities should be investigated with detailed USG and, if necessary, MRI. Our four patients had isolated DWC, and neurological examination findings were within normal limits in all of them. The image of the patient diagnosed with the DWC is shown in Figure-1. The mortality rate was 100% in all our patients with additional findings. In the study of Scarlet et al., the live birth rate was 87% in the group with isolated pf, while the live birth rate was only 52% in the non-isolated group (8). Four of our eight patients with DWC, whose postnatal results we could follow up (the other four patients who could not be followed up were terminated and no autopsy was performed, and one was intra-uterine exitus), had an extra anomaly. Has et al. detected DWC in 14 patients in a series of 78 patients; 3 of them had additional cardiac abnormalities (9). In our series, two patients had congenital heart abnormalities, and both patients died after cardiovascular surgery in the postnatal period. Therefore, we think that cardiac abnormalities have a significant determinant role in neonatal prognosis.

There are contradictory results in the literature regarding the spectrum of isolated DWC. In the series of six patients by Guibad et al., three patients had normal neurological findings, but one patient also had partial trisomy seven and moderate-to-severe neurological impairment (10). In our study, four patients had isolated DWC, and all had average neurological results. If the genetic examination results are average in the isolated group, the prognosis is mostly good, and it is necessary to inform the family.

Dror et al. observed that the prognosis was good in patients with isolated MSM in their series of 29 patients, and they found that only the first step period was delayed (10). Four of our patients were diagnosed with isolated MSM, and all of them had an average neurological examination. MR image of a patient diagnosed with isolated MSM is shown in Figure-2. We did not detect the first-step delay in any of our patients with isolated MSM pathology.

Prenatal diagnosis of rhomboencephalosynapsis is rare and generally results in termination, or various degrees of neurological abnormalities were found in surviving infants (11, 12). Poretti et al. In their case series consisting of 5 patients in 2009, they reported that two patients had normal neurological development (13). One of our patients was diagnosed with isolated partial rhomboencephalosynapsis, and no additional neurological anomaly was detected. Prenatal and postnatal MR images of the patient diagnosed with rhomboencephalosynapsis are shown in Figure-3.

One of our patients was diagnosed with unilateral cerebellar hypoplasia (UCH) and had a balance problem in the postnatal period. The prenatal and postnatal view of the patient with UCH is shown in Figure-4.

Massoud et al. reported that the neurological prognosis is good if UCH is not associated with posterior fossa abnormalities, hemangioma, arterial abnormalities, cardiac abnormalities/aortic coarctation, eye abnormalities syndrome (PHACE syndrome), or an infection (14). The accompanying balance problem in our patient may be due to Vermian hypoplasia.

Diagnosis of PFA abnormalities is complex, and it is a condition that cannot be predicted clearly which patients will have a good prognosis and which will have a bad prognosis. The prognosis is not affected by maternal and fetal factors and allows the recognition of additional accompanying abnormalities. Except for one case, fetal MRI findings were similar to fetal ultrasound findings. (We thought that one patient had DWC Anomaly on USG, but fetal MR detected UCH.) Fetal MRI is an imaging method that can provide retrospective examination and research, especially in pregnancies with poor prognoses.

Conclusion

This study includes the long-term results of fetal posterior fossa anomalies and we think that it can provide information to experts working in this field. Therefore, we can say that the prognosis of the child will be good if there is only MCM in fetal MRI and there is no extra anomaly. However, there may be extra anomalies that can only

be detected in the postpartum period. Therefore, especially USG and fetal MRI should be performed together for long-term outcome prediction in fetuses diagnosed with fetal PFA.

Acknowledgement
This article was presented at the congress of TMFTP in 2021.

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Table 1. Demographic characteristics of patients	
Demographic data	
Maternal age (years) [mean(min-max)]	23.68 (21-43)
Pregnancy age at MRI (years) [mean(min-max)]	26.34 (19-36)
Maternal additional disease	2 hypothyroidism, 1 GDM
Multiple gestations (n)	3
ART (n)	2
Birth age (weeks) [mean(min-max)]	26.27 (23-27)

Birth age (weeks, except terminations) [mean(min-max)]	36.97 (35-40),
Birth weight (grams, except terminations) [mean(min-max)]	2911.76 (425-4100)
The age range of children (year) (min-max)	1-8
*Mean (min-max) ART: Assisted reproduction technology, GDM: Gestational diabetes mellitus, n: Number	

Table 2. Clinical outcome of posterior fossa abnormalities

	Diagnosis	Number of patients	Pregnancy Outcome	Current status	Neurological Outcome	Additional anomaly
Primer posterior fossa abnormalities	MCM	4	Live birth	Living	Normal	None
	Rhomboencephalosynapsis	2	Live birth	Living	Normal	Esophageal atresia
			Preterm live birth	Postpartum exitus		Hydrocephalus
	DWC	14	9 Live birth	7 Living	3 Abnormal	1-Hydrocephalus 2-Cerebellar hypoplasia
					4 Normal	
				2 Postpartum exitus		1-DORV 2-Diastometamyelia + interrupted aorta
			4: TOP 1: IUEX			
Seconder posterior fossa abnormalities	NTD	2	Live birth	Arnold-Chiari malformation	Normal	
			Iniencephaly postpartum exitus			

DWM: Dandy-Walker Continuum, MCM: Mega cisterna magna, NTD: Neural-tube defect, TOP: Termination of pregnancy, IUEX: Intrauterine exitus, DORV: Double outlet right ventricule

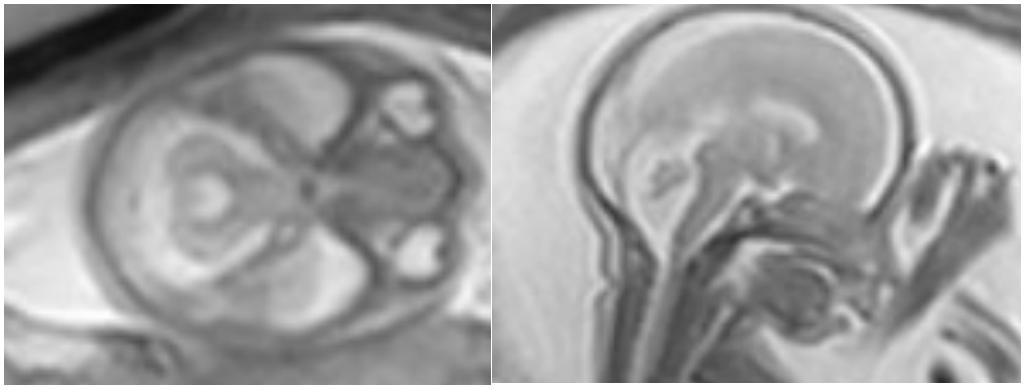

Figure 1. Dandy Walker Continuum

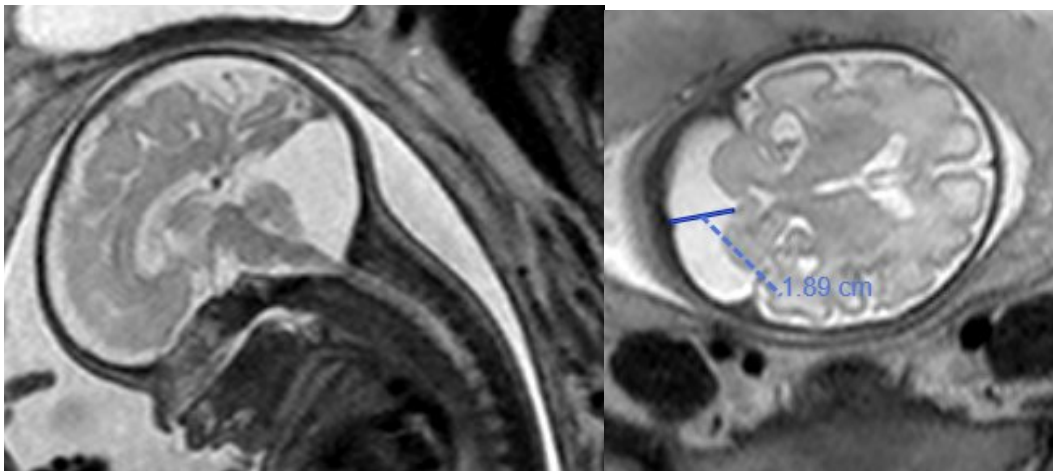

Figure 2. Mega cisterna magna

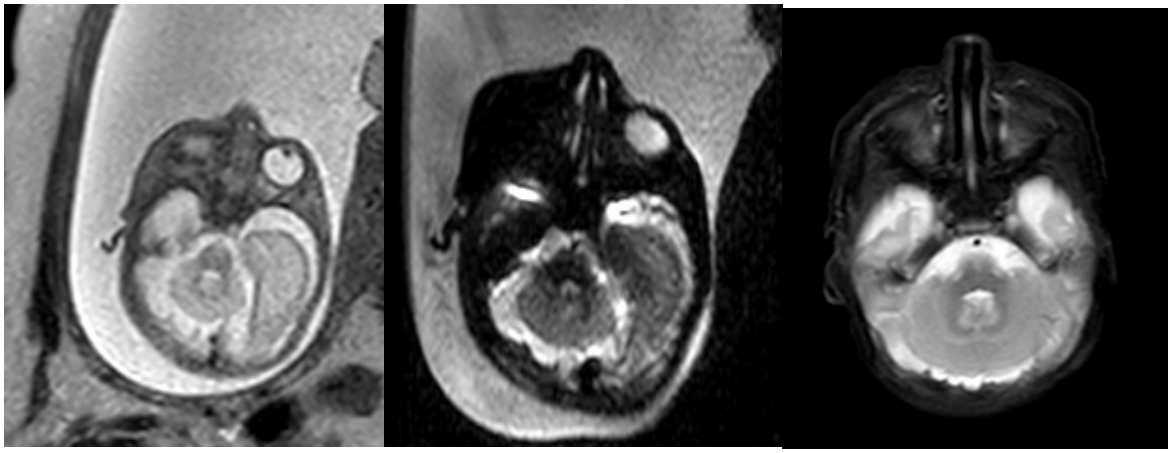

Figure 3. Rhomboencephalosynapsis

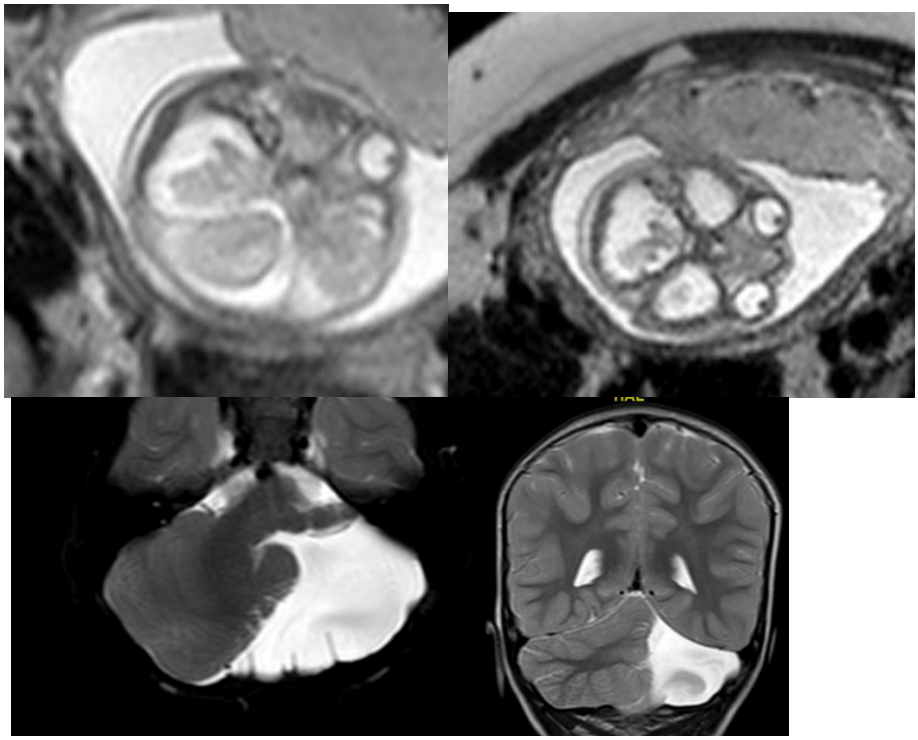

Figure 4. Unilateral cerebellar hypoplasia