Long-term outcomes of fetal posterior fossa abnormalities diagnosed with fetal magnetic resonance imaging

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Abstract

Objective: The diagnosis of posterior fossa abnormalities (PFA) in the intrauterine period and association with pregnancy outcomes are still controversial. PFA is generally referred to 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 in patients diagnosed with PFA between January 2013 and September 2020 in a tertiary perinatology clinic. All patients underwent routine second-trimester ultrasound screening and definitive diagnosis was made by fetal magnetic resonance imaging (MRI) in the presence of a suspected anomaly.

Results: There were 164 fetal MRIs for fetal abnormalities during the study period and 22 (13.4%) were diagnosed with a PFA on fetal MRI. Indications for fetal MRI included four (18%) with Mega Cisterna Magna, two (9.1%) with rhomboencephalosynapsis, and thirteen (59.1%) with Vermian Hypoplasia-Dandy-Walker variant. Two patients, with neural tube defects and lumbosacral neural-tube defect are still alive. However, iniencephaly was detected in last patient who died in the postnatal period.

Conclusion: Diagnosis of PFA abnormalities is complex, and the prognosis in PFA is often unclear. 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. (J Turk Ger Gynecol Assoc 2023; 24: 28-32)

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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 cerebellar anomaly on child development should be investigated.

The diagnosis of posterior fossa abnormalities (PFA) in the intrauterine period and the association with pregnancy outcomes are still controversial. PFA is usually referred to a maternal-fetal medicine specialist. The incidence of PFA is approximately 1:5,000 in live births (4). PFA is generally divided into two groups - primary and secondary. Primary PFA is further subdivided into 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 regions (6). Basic ultra-sonographic (USG) evaluation of PFA is usually performed in the second trimester transabdominally using a transverse

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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 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. The sagittal expansion of the PF is usually associated with an elevated position of the tentorium and its occipital insertion into the torcular herophili region. In PFA, fetal magnetic resonance imaging (MRI) may be used when the diagnosis cannot be made by USG examination, or to confirm the diagnosis. It has been reported that potential USG 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 was to analyze the neurological outcomes of children diagnosed with PFA prenatally using fetal MRI.

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 Faculty of Medicine, Department of Obstetrics and Gynecology. All patients underwent routine second-trimester USG screening, and the definitive diagnosis was made by fetal MRI (Philips Ingenia 1.5 Tesla), in the presence of a suspected anomaly. In addition, patients diagnosed with any anomaly in another center and referred were also included in the study. USG examination of all patients was performed with Voluson 8 (GE Healthcare Ultrasound, Milwaukee, WI, USA), using a 4-8 Mhz convex transabdominal transducer, and a 5-9 Mhz transvaginal transducer. The records of patients who underwent postnatal MRI examination were also reviewed.

Demographic data of patients diagnosed with PFA were analyzed from hospital records, and an experienced perinatologist reviewed USG images. The images were compared with fetal MRI recordings. Amniocentesis was performed in mothers who accepted amniocentesis. When an abnormality was detected, information about the prognosis was given by the perinatology specialist. Termination of pregnancy (TOP) was performed according to parental choice. In Turkey, there is no upper limit for gestational week for TOP 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 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 neurological developmental results of the fetuses were recorded. Detailed neurological and developmental examination could not be performed because surviving babies were often resident far from the study center. Information about vision, speech and motor function of the patients was obtained from the parents by telephone. Mechanical ventilator dependence was investigated.

This study was conducted following approval by Ankara University Cebeci Hospital Clinical Research Ethics Committee (approval number: İ06-355-22) and written informed consent was obtained from all participants.

Statistical analysis

IBM SPSS, version 26.0 for Windows (SPSS Inc., Chicago, IL, USA) was used for statistical evaluation of 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 standard deviation, and categorical factors were summarized using proportions.

Results

Between 2013 and 2020, 164 fetal MRIs were performed for fetal abnormalities, and 22 (13.4%) fetuses were diagnosed with a PFA on fetal MRI. The mean age of the patients was 23.68 (21-43) weeks. The mean gestational 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 (425-4,100) grams. The ages of the surviving participants ranged from 1 to 8 years. The demographics of the patients are given in Table 1.

In terms of indications for fetal MRI, four (18%) were diagnosed with Mega Cisterna Magna (MSM), and the neurological examination of all patients was within normal limits. Two (9.1%) 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 (63.6%) were diagnosed with DWC, and four of them were terminated. One pregnant woman presented with abruptio placentae and USG revealed that the fetus had no cardiac activity. Two (9.1%) died in the postnatal period, and both had additional abnormalities. One had a double out right ventricle, and the other had interrupted aorta and diastematomyelia. There were six

(27.3%) surviving infants and two of them had additional abnormalities besides neurological abnormalities. One had cerebellar hypoplasia, and the baby had speech delay and balance problems. The other had 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. Two patients with neural tube defects and lumbosacral neural-tube defect are still alive. However, iniencephaly was detected in the last patient who died in the postnatal period. Abnormalities of fetus and long-term results of survivors are shown in Table 2.

Table 1. Demographic characteristics of patients

Demographic data	
Maternal age (years) [mean (minimum-maximum)]	23.68 (21-43)
Pregnancy age at MRI (years) [mean (minimum-maximum)]	26.34 (19-36)
Maternal additional disease	2 hypothyroidism, 1 GDM
Multiple gestations (n)	3
ART (n)	2
Birth age (weeks) [mean (minimum-maximum)]	26.27 (23-27)
Birth age (weeks, except terminations) [mean (minimum-maximum)]	36.97 (35-40)
Birth weight (grams, except terminations) [mean (minimum-maximum)]	2911.76 (425-4100)
The age range of children (years)	1-8

Mean (minimum-maximum). ART: Assisted reproduction technology, GDM: Gestational diabetes mellitus, n: Number, MRI: Magnetic resonance imaging

Discussion

Fetal MRI is generally preferred in patients whose definitive diagnosis cannot be made after second-trimester USG, and it reveals additional undetected findings when compared with USG at a rate of 60% (8). In this study, fetal MRI was performed between 26 and 34 weeks of gestation, in line with the literature. One of the main results 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. 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 with additional findings. In the study of Scarlet et al. (8), the live birth rate was 87% in the group with isolated PFA, while the live birth rate was only 52% in the non-isolated group. Four of our eight patients with DWC had postnatal results available for follow up while in the other four, all had died with most being terminated without autopsy while one was intra-uterine exitus with an extra anomaly. Has et al. (9) detected DWC in 14 patients in a series of 78 patients and three of them had additional cardiac abnormalities. In our series, two patients had congenital heart abnormalities, and both patients died after cardiovascular surgery in the postnatal period. Therefore, it appears that cardiac abnormalities have a significant determinant role in neonatal prognosis.

Tablo 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

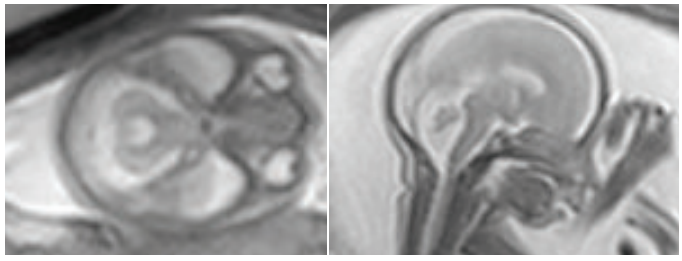

Figure 1. Dandy Walker continuum

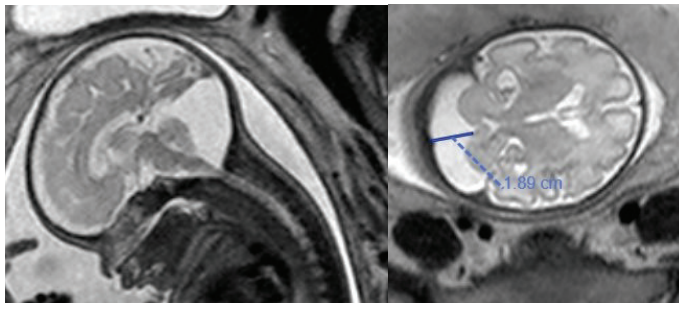

Figure 2. Mega cisterna magna

There are contradictory results in the literature regarding the spectrum of isolated DWC. In the series of six patients by Guibad et al. (10), three patients had normal neurological findings, but one patient also had partial trisomy seven and moderate-to-severe neurological impairment. 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 the family should be so informed.

Dror et al. (10) reported 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. Four of our patients were diagnosed with isolated MSM, and all of them had average neurological examination findings. An MRI 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, while various degrees of neurological abnormalities were found in surviving infants (11,12). Poretti et al. (13) in a case series of five patients from 2009 reported that two patients had normal neurological development. One of our patients was diagnosed with isolated partial rhomboencephalosynapsis, and no additional neurological anomaly was detected. Prenatal and postnatal MRIs of this patient diagnosed with rhomboencephalosynapsis are shown in Figure 3.

One patient 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. (14) reported that neurological prognosis

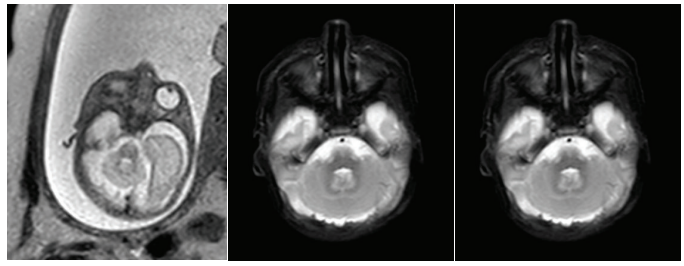

Figure 3. Rhomboencephalosynapsis

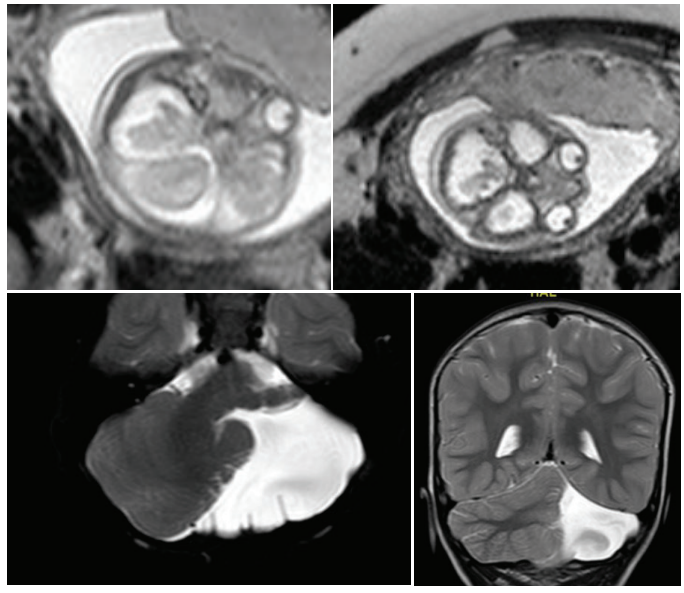

Figure 4. Unilateral cerebellar hypoplasia

is good if UCH is not associated with PFA, hemangioma, arterial abnormalities, cardiac abnormalities/aortic coarctation, eye abnormalities syndrome (PHACE syndrome), or an infection. The accompanying balance problem in our patient may be due to Vermian hypoplasia.

Diagnosis of PFA abnormalities is complex, and patient prognosis is unclear. The prognosis is not affected by maternal and fetal factors. Fetal MRI allows the recognition of additional accompanying abnormalities. Except for one case, fetal MRI findings were similar to fetal USG findings, in which one patient was diagnosed with DWC anomaly on USG, but fetal MR indicated UCH. Fetal MRI is an imaging method that can provide retrospective examination and research, especially in pregnancies with poor prognoses.

Conclusion

This study reports the long-term results of fetal PFA and we hope that these will be of interest to experts working in this field, given the limited data available. Our findings suggest that the prognosis of the child will be good if there is isolated MCM on fetal MRI and there is no extra anomaly. However, there may be extra anomalies that can only be detected in the postpartum

period. Therefore, we believe that USG and fetal MRI should be performed together for long-term outcome prediction in fetuses diagnosed with fetal PFA.

Ethics Committee Approval: *This study was conducted following approval by Ankara University Cebeci Hospital Clinical Research Ethics Committee (approval number: İ06-355-22).*

Informed Consent: *Written informed consent was obtained from all participants.*

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