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PRENATAL AND POSTNATAL EVALUATION OF DANDY-WALKER MALFORMATION : A CASE REPORT

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We described a case of Dandy-Walker syndrome, that was detected in utero and was evaluated and monitored postnatally. The prenatal examination by echography and MRI revealed a large posterior fossa cyst with splaying of the cerebellar hemispheres. The male infant was born at 40 weeks of gestation, weighing 3430 g with no special problems. Although the infant presented transient tachypnea immediately after birth, he has had no neurological symptoms. Echography, computed tomography (CT) and magnetic resonance imaging (MRI) performed after birth revealed an enlargement of the fourth ventricle but not of the lateral ventricles, and dysgenesis of the cerebellar vermis. Circumference of the head was normal and no extracranial anomalies except for the left simian crease were noted. Chromosomal analysis showed normal male pattern, 46, XY. He has been carefully followed up with no neurosurgical treatment. He has showed normal growth and development, and no hydrocephalus, as of 3 months of his age.

Key words: Dandy-Walker malformation/the value of prenatal diagnosis / prenatal echography / postnatal evaluation

Dandy-Walker malformation (DWM) is a congenital cranial anomaly, characterized by complete or partial absence of the cerebellar vermis, cystic dilatation of the fourth ventricle, and, often, hydrocephalus. DWM is often associated with other central nervous system and extracranial anomalies. Chromosomal abnormality is occasionally noted. The incidence of DWM is considered approximately 1/25,000 to 30,000 (1). Intrauterine diagnosis of such a disease will be of help early treatment after birth, such as neurosurgical treatment to reduce the intracranial pressure, if necessary. We report here a male infant of DWM, who was prenatally detected by ultrasonography and postnatally evaluated and monitored. He has showed normal growth and development with no specific treatment for a few months, up to now.

CASE PRESENTATION

A 32-year-old woman was referred to our hospital, because of an abnormality in ultrasonographic examination at 31 weeks of gestation. A large echo free space in fetal cranium was noted. Further detailed examination of echographic scan revealed a large free space in the posterior fossa with splaying in the cerebellar hemispheres of the fetus, as shown in Figs 1A and 1B. The sagittal scan demonstrated cystic dilatation of the fourth ventricle, dysgenesis of the cerebellar vermis, and elevation of the tentorium. It was strongly suspected that this fetus had DWM. Magnetic resonance imaging (MRI) of the pregnant woman performed at the 34 weeks of gestation also revealed the similar findings to that of the echographic scan, being consistent with the diagnosis of DWM, as shown in Fig 2A. No enlargement of the ventricles were noted during the pregnancy as shown in Fig 2B and the pregnancy was continued.

The male infant was delivered naturally at 40 weeks of gestation, weighing 3430 g with no asphyxia (Apgar score 8 and 9 at 1 and 5 minutes, respectively). At birth, his length was 49 cm; circumference of the head, 34.5 cm; circumference of the chest, 32.5 cm. No abnormal neurological signs were noted. The routine laboratory tests revealed no special abnormalities as shown in Table 1: CRP, $<$0.2 mg/dl; IgM, 12 mg/dl and so on. Although mild tachypnea was noted for the first 2 days, it disappeared by only oxygen therapy of FiO_2, 30 %. Chromosomal examination revealed a normal male karyotype, 46, XY. No other minor anomalies were noted except for the left simian crease.

In ultrasonography , CT scan and MRI examinations, an enlargement in the fourth ventricle was noted but not in the lateral ventricles, as shown in Figs 3A and 3B. The ultrasound examinations were carried out at 7, 28 and 57 days after birth, and the size of lateral ventricles has been normal. MRI revealed mild thickness of the corpus callosum other than the above abnormalities as showen in Fig 3A. With the above clinical findings, we are following up on his growth and development as well as morphological tests in the periodic medical check.

Table 1. Laboratory findings at birth

Periferal blood		Blood biochemical examinations	
WBC	17300 /mm^3	TP	6.4 g/dl
RBC	5.49x10^6 /mm^3	Alb	4.0 g/dl
Hb	18.9 g/dl	T.bil	1.8 mg/dl
Ht	56.4 %	GOT	45 IU/l
Plt	192x10^3 /mm^3	GPT	17 IU/l
		LDH	380 IU/l
		CK	333 IU/l
Urinalysis		BUN	10 mg/dl
no particular abnormalities		Crea	0.7 mg/dl
		Na	139 mEq/l
Heart echography		K	4.1 mEq/l
no particular abnormalities		Cl	108 mEq/l
		CRP	<0.2 mg/dl
Abdominal echography		IgG	1474 mg/dl
no particular abnormalities		IgA	<62 mg/dl
		IgM	12 mg/dl
Chromosomal analysis			
46, XY			

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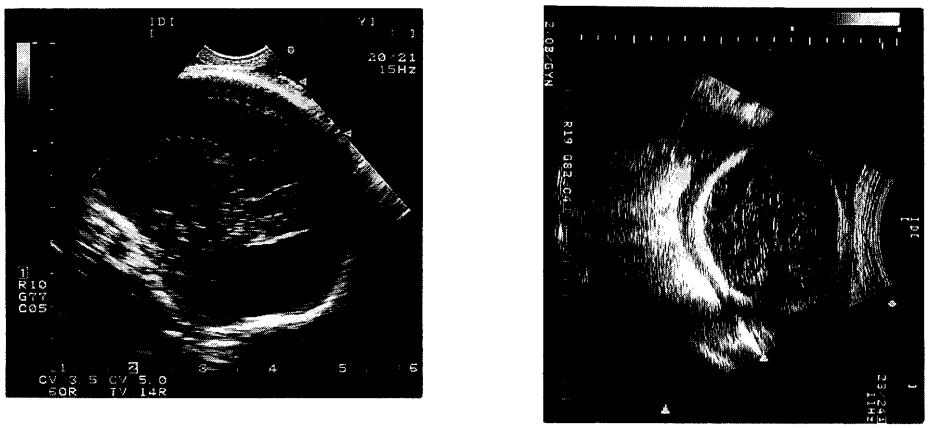

Fig. 1. Prenatal ultrasonography findings at 31weeks of gestation. A, Sagittal scan. A large free space in the posterior fossa cyst, dysgenesis of the cerebellar vermis and elevation of the tentorium were noted. B, Axial scan. A cystic dilatation of the fourth ventricle with splaying in the cerebellar hemispheres was noted.

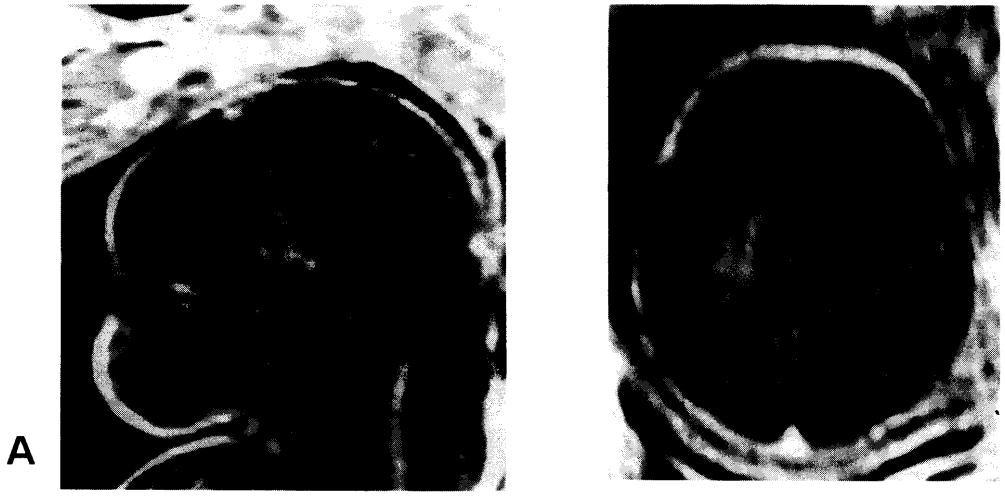

Fig. 2. T1 weighted image (TR 50msec, TE 4.1msec) of 1.5 Tesla MRI on 34 weeks of gestation. A, Sagittal images. Cystic dilatation of the fourth ventricle, dysgenesis of the cerebellar vermis and elevation of the tentorium were noted. B, Axial images. No enlargement of lateral ventricles were noted.

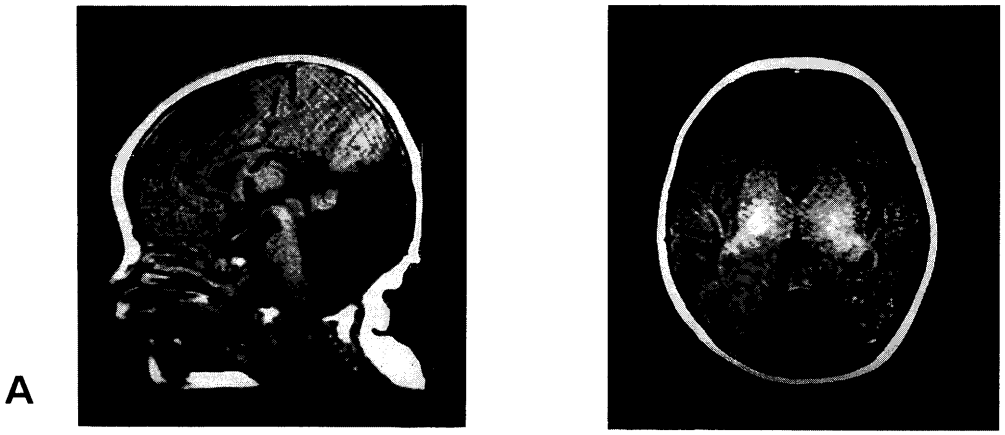

Fig. 3. T1 weighted image (TR 400msec, TE 14msec) of 1.5 Tesla MRI on 50 days after birth. A, Sagittal images. Cystic dilatation of the fourth ventricle, elevation of the tentorium and thickness of the corpus callosum were noted. The rudimentary incomplete vermis now becomes recognizable. B, Axial images. No enlargement of the lateral ventricles nor migration disorders of the cerebral cortex were noted.

DISCUSSION

According to the Raybaud's original classification (2), cystic lesions in the posterior fossa were classified into DWM, Dandy-Walker variant and retrocerebeller cyst. The classical DWM is defined with the triad of cystic dilatation of fourth ventricle, complete or partial agenesis of cerebellar vermis, and an enlarged posterior fossa, associated with displacement of the tentorium, torcular and lateral sinus. Dandy-Walker variant is distinguished from DWM by milder partial agenesis of cerebeller vermis and no signs of an enlarged posterior fossa. This case was a typical DWM.

DWM is considered to be caused by an abnormal fetal development of the encephalic midline structures before the 6 or 7 weeks of gestation. Approximately 70% of DWM patients have other CNS anomalies such as corpus callosum dysgenesis, subependymal neuronal heterotopia, polymicrogria, agyria, schizencephaly, lipoma of the corpus callosum, encephalocele, or lumbosacral meningocele (3-6). This infant had mild dysgenesis of the corpus callosum. Hydrocephalus often occurrs in around 75% of cases of classical DWM. Occasionally, hydrocephalus occurs a few months after birth, even if no sign for it at birth. Our case has shown no such manifestations for a few months. At least 25% of DWM patients have also been reported to be associated with extracranial abnormalities, such as low-set ear, cleft lip, cleft palate, facial angiomas, polydactyly, synodactyly, Klippel-Feil syndrome, or Cornellia de Lange syndrome. DWM was also associated with several genetic disorders, including autosomal recessive diseases, Meckel-Gruber syndrome, Warburg syndrome or X-linked Aicardi syndrome. Further, karyotypic abnormalities have also been reported, such as duplications of 5p, 8p, or 8q; trisomy of 9, 13 and 18; and triploidy (7). Intrauterine exposure to rubella, cytomegalovirus, toxoplasma, warfarin, or alcohol were also reported (5,7,8). In our case, no such findings or history were noted.

Some people would have a question on examination of CT scan for the diagnosis of DWM, because of less diagnostic significance and irradiation to fetuses (4). We believe, however, that the findings of CT scan are informative from the images. The CT features are reported : (a) partially or, less commonly, complete absence of the vermis, (b) cystic dilatation of the fourth ventricle that impringes on the occipital bone, (c) an enlarged posterior fossa with elevated tentorium (4). In our case, almost complete absence of cerebellar vermis, cystic dilatation of the fourth ventricle and an enlarged posterior fossa were observed. Furthermore, the MRI may provide a more sufficient evidence on the posterior fossa cysts, especially in case of rotation or upward displacement of the cerebellar vermis, although the cyst or cistern cannot be evaluated in detail by MRI (9). We are going to further evaluation by cine-MRI on these subjects in this patient.

In this case, DMW was prenatally indicated, so that the accurate evaluation and proper care could be performed immediately after birth. It was reported that 20 to 60% (average, around 38.6%) of DWM patients had normal intelligence (1,4,6,7). In the patients whose cerebral cortex is not affected or decreased, the mental prognosis tends to be good (10). We monitored enlargement of ventricles of this patient during pregnancy and after birth and decided observation without neurosurgical treatment. Choice of the mode of delivery might also affect the outcome. Prenatal detection will be of help in early evaluation of severity, other complicated anomalies, karyotypic abnormality, genetic disorders associated, or even in decision of mode of delivery. The value of prenatal diagnosis by echography has been emphasized by some obstetricians (10) and radiologists (5-7). This is a case report in that the value of prenatal detection and postnatal concrete evaluations were discussed mainly in view of pediatritians.

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