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Prenatal diagnosis of diastematomyelia: presentation of eight cases and review of the literature

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KEYWORDS: diastematomyelia; diplomyelia; prenatal diagnosis; split cord malformation

ABSTRACT

Objectives *Diastematomyelia is a rare form of spinal dysraphism. We present eight cases of diastematomyelia diagnosed prenatally in our institution as well as a review of the literature in order to determine the prognosis of isolated cases of this very unusual condition.*

Methods *Records of fetuses with diastematomyelia diagnosed in our institution between January 2000 and June 2005 were collected. All liveborn fetuses were examined by a pediatric neurologist. Pre- and postnatal data were analyzed. A search was then conducted using PubMed to review previously reported cases in the literature.*

Results *Eight cases of diastematomyelia were diagnosed during the study interval. The mean (range) gestational age at diagnosis was 21 (13–25) weeks. The main sonographic findings were widening of the spinal canal in the coronal view and an additional echogenic focus in the posterior part of the spinal column in the axial view. The diagnosis of associated open spina bifida was made in one fetus with elevated levels of amniotic fluid α-fetoprotein (AF-AFP) and acetylcholinesterase (AF-AChE) and the pregnancy was terminated. The other seven cases of diastematomyelia had normal levels of AF-AFP and AF-AChE and were considered isolated. One pregnancy miscarried spontaneously 1 week following amniocentesis and the remaining six were delivered at term. Review of the literature revealed 14 reports involving 26 cases of prenatally diagnosed diastematomyelia. Twelve cases had normal biochemistry and/or no additional abnormalities and all had a favorable outcome.*

Conclusions *When diastematomyelia is not associated with other spinal anomalies, the prognosis is favorable. Prenatal diagnosis is generally made in the second trimester but sonographic signs may be recognized as early as the first trimester. Intrauterine diagnosis of diastematomyelia should facilitate appropriate management of affected cases. Copyright © 2007 ISUOG. Published by John Wiley & Sons, Ltd.*

INTRODUCTION

Diastematomyelia is a spinal abnormality in which the spinal cord is divided into two columns by a cartilaginous or osseous spur. The synonyms of diastematomyelia are split cord malformation and diplomyelia. In most cases, lesions are located on the lower thoracic and upper lumbar parts of the spine[1]. The most striking ultrasonographic findings of diastematomyelia are a widening of the spinal canal in the coronal plane and an echogenic spur traversing the spinal canal in the axial plane. Diastematomyelia may be isolated or it may be associated with other spinal anomalies such as spina bifida, kyphoscoliosis, butterfly vertebra or hemivertebra.

METHODS

Data were collected from fetuses with diastematomyelia diagnosed in our institution between January 2000 and June 2005. The ultrasonographic criteria for diagnosis were a widening of the spinal canal in the coronal view, an additional echogenic focus crossing the spinal canal between the anterior and the posterior walls in the axial view and intact skin and soft tissues overlying the affected spinal segment (Figures 1 and 2). We also evaluated amniotic fluid acetylcholinesterase (AF-AChE) and amniotic fluid α-fetoprotein (AF-AFP) levels for the possibility of associated open spina bifida. Although the first-trimester screening tests were normal for all patients, we also performed a chromosomal analysis of the amniotic fluid taken during the AF-AChE and AF-AFP analysis.

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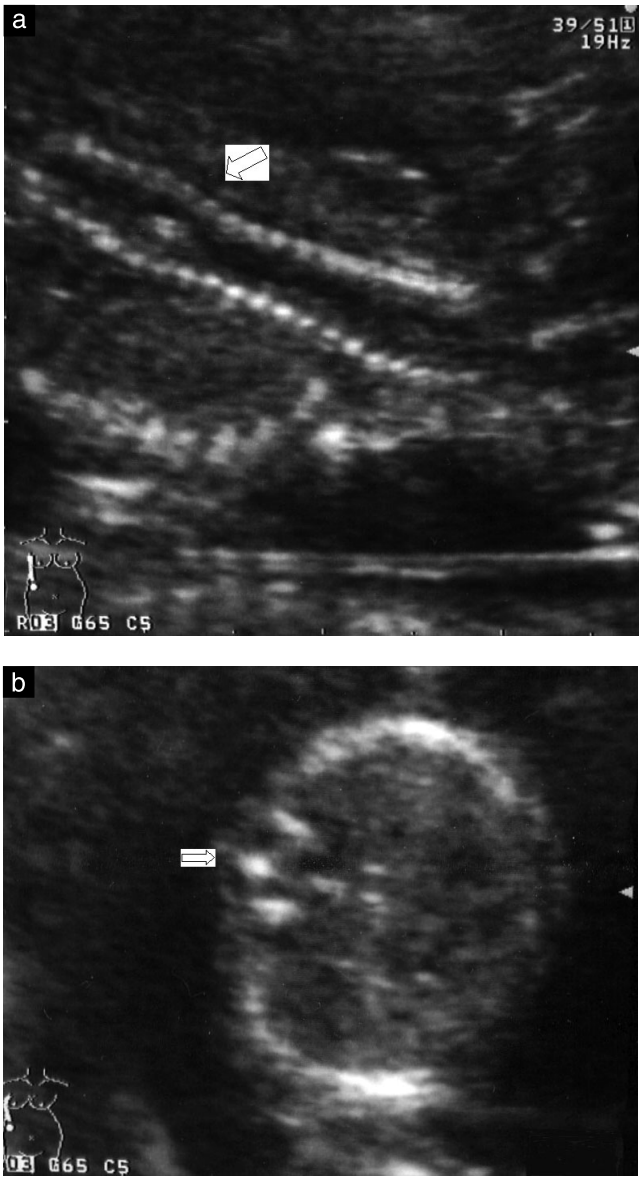

Figure 1 Case 3. Coronal and axial ultrasound images of vertebrae at 13 weeks' gestation showing diagnostic signs of diastematomyelia: (a) widening of the spinal canal in coronal section (arrow) and (b) additional echogenic focus in the posterior part of the vertebral column in axial section (arrow).

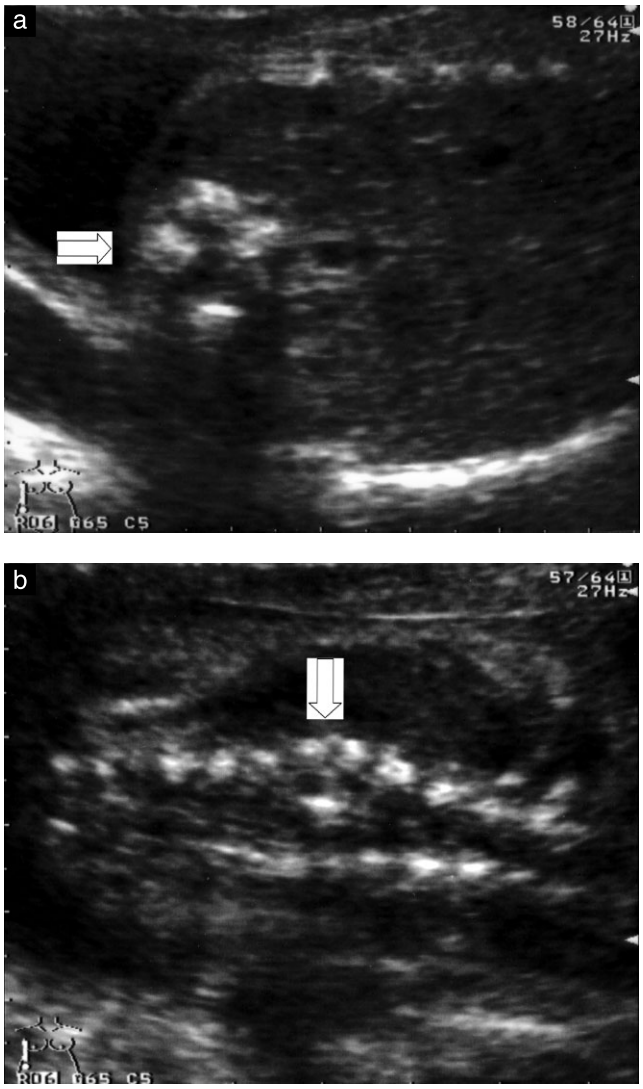

Figure 2 Case 5. Ultrasound images at 23 weeks' gestation showing an extra echogenic focus in axial (a) and coronal (b) sections (arrows).

Obstetric data of the women were evaluated and all liveborn fetuses were examined by a pediatric neurologist.

A review of the literature was carried out using the PubMed database, with the search terms diastematomyelia, split cord malformation, diplomyelia and prenatal diagnosis; the latest search was done in May 2006.

RESULTS

During the study period, eight cases of diastematomyelia were diagnosed in 27 085 women (0.30‰). The mean ± SD gestational age at diagnosis was 21.1 ± 3.9 weeks. All women consented to undergo amniocentesis for chromosomal, AF-AChE and AF-AFP analysis. Karyotype analyses were normal in all cases with only a single case with open spina bifida testing positive for AF-AChE and AF-AFP. This pregnancy was terminated following the amniocentesis. Autopsy revealed open spina bifida and diastematomyelia at the low thoracic segment. One fetus miscarried spontaneously 1 week after the amniocentesis at 24 weeks' gestation. Focal pigmentation and hair were observed on the skin overlying the lumbar part of the spine. The family did not consent to an autopsy.

The remaining six fetuses were examined for possible additional abnormalities; one had a single umbilical artery. All six women delivered at term. Neurological examination of the babies was normal, however they all had dimples, focal pigmentation and/or hair over the affected spinal segment (Figure 3). Their spinal anatomy was evaluated using magnetic resonance imaging (MRI), which in all cases revealed diastematomyelia without any other spinal malformation (Figure 4). The prenatal sonographic findings and obstetric outcomes are shown in Table 1.

Fourteen papers reporting a total of 26 cases of the prenatal diagnosis of diastematomyelia were pooled and

Table 1 Prenatal sonographic findings and postnatal outcomes of fetuses with diastematomyelia

Case	*MA (years)*	*GA at diagnosis (weeks)*	*Indication*	*AF-AFP, AF-AChE status*	*Additional sonographic findings*	*Outcome*	*Examination after miscarriage/TOP or prognosis after birth*
1	22	23	Spina bifida	Negative	None	Spontaneous miscarriage 1 week after amniocentesis	Focal pigmentation and hair bunch observed in the skin overlying the lumbar part of the spine*
2	21	18	Spina bifida	Positive	Open spina bifida in low thoracic region	TOP	Autopsy revealed an open spina bifida and diastematomyelia at the low thoracic segment
3	22	13	First-trimester screening	Negative	None	Uneventful pregnancy and vaginal delivery at term	Thoracic diastematomyelia (T3–4); operation at age of 5 months owing to tethered cord. No neurological sequelae at age of 1 year
4	30	23	Spina bifida	Negative	None	Uneventful pregnancy and vaginal delivery at term	Lumbar diastematomyelia (L2); asymptomatic at age of 6 months
5	26	23	Routine	Negative	None	Uneventful pregnancy and Cesarean section at term	Thoracic diastematomyelia (T10–12); asymptomatic at age of 20 months
6	34	24	Spina bifida	Negative	None	Uneventful pregnancy and Cesarean section at term	Sacral diastematomyelia (S2–3); asymptomatic at age of 1 year
7	30	20	Spina bifida	Negative	None	Uneventful pregnancy and vaginal delivery at term	Thoracic diastematomyelia (T11–12); operation at age of 16 months owing to tethered cord. No neurological sequelae at age of 3 years
8	25	25	Spina bifida	Negative	SUA	Uneventful pregnancy and Cesarean section at term	Thoracic diastematomyelia (T7–8); asymptomatic at age of 18 months

*The family did not authorize an autopsy. AF-AChE, amniotic fluid acetylcholinesterase; AF-AFP, amniotic fluid alpha-fetoprotein; GA, gestational age; MA, maternal age; SUA, single umbilical artery; TOP, termination of pregnancy.

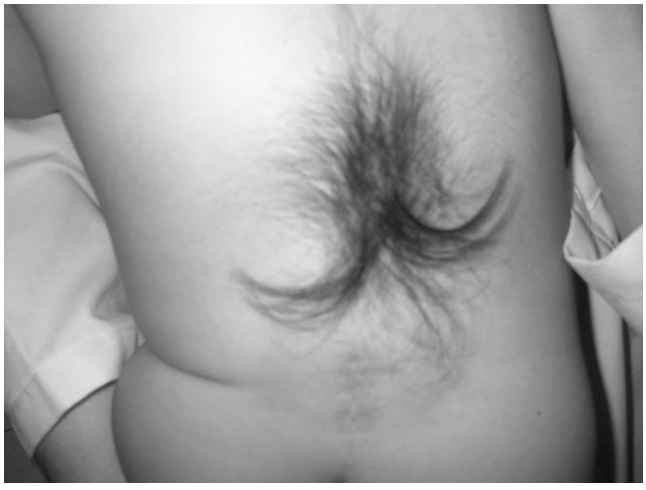

Figure 3 Photograph of hair, pigmentation and dimples that were present in the skin overlying the affected segment of the vertebral column.

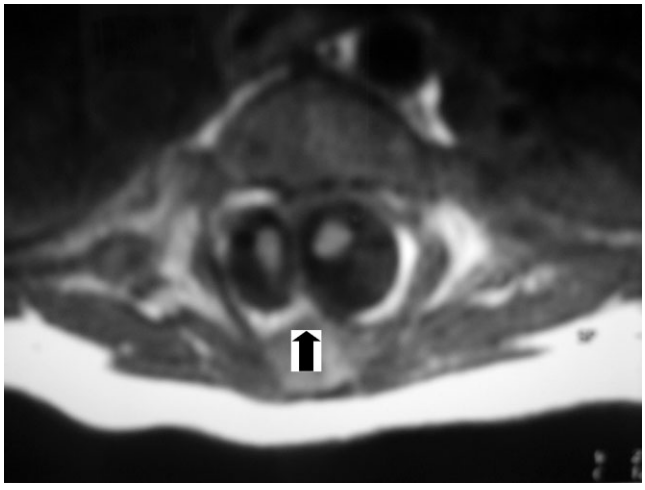

Figure 4 Postnatal T1-weighted magnetic resonance image showing diastematomyelia: the arrow shows the septum dividing the spinal canal from the anterior wall to the posterior wall.

the results are shown in Table 2. The most relevant finding was that the 12 fetuses with normal biochemistry and/or where diastematomyelia was the only abnormality had a favorable prognosis.

DISCUSSION

Diastematomyelia is a rare abnormality of the neural tube and it is usually associated with other spinal

Table 2 Review of previously reported cases of diastematomyelia

Reference	*MA (years)*	*GA at diagnosis (weeks)*	*Indication*	*Additional sonographic findings*	*AF-AFP, AF-AChE and MS-AFP status*	*Outcome*
Suma *et al.*[1]	28	26	Triplet pregnancy (one affected)	None	?	Liveborn; asymptomatic (all three babies)
Williams and Barth[11]	26	23	Hydrocephaly	Myelomeningocele	Elevated MS-AFP	TOP
Winter *et al.*[12]	18	15	Routine	Butterfly vertebra	Normal MS-AFP and AF-AFP	Liveborn; small foot, short leg, butterfly vertebra, spinal surgery
	21	20	Routine	None	Normal MS-AFP	Liveborn; butterfly vertebra, hemivertebra, asymptomatic
	19	16	Routine	None	Normal MS-AFP	TOP
	23	18	Routine	None	Normal MS-AFP and AF-AFP	TOP
Caspi *et al.*[2]	29	19	Elevated MS-AFP	Open spina bifida	Elevated MS-AFP	TOP
Gubbels *et al.*[10]	26	17	Maternal acyclovir user	None	Normal MS-AFP	TOP
Pachi *et al.*[8]	30	20	Maternal diabetes	Scoliosis	Normal MS-AFP and AF-AFP	TOP
Boulot *et al.*[9]	25	33	Abnormal fetal spine	Spina bifida, kyphosis	Normal AF-AChE	TOP
Anderson *et al.*[5]	?	18	Routine	None	?	TOP
	?	?	Routine	None	?	Fetal demise at 25 weeks
	?	20	Routine	None	?	Liveborn; spinal surgery
	?	?	Routine	None	?	Liveborn; asymptomatic
	?	19	Routine	Meningocele	Normal AF-AFP	TOP
Sepulveda *et al.*[4]	28	21	Routine	None	Normal MS-AFP	Liveborn; asymptomatic
	32	21	Routine	None	Normal MS-AFP	Liveborn; asymptomatic
Allen and Silverman[3]	21	28	Routine	None	Normal MS-AFP and AF-AChE	Liveborn; spinal surgery
	31	17	Abnormal fetal spine	None	Normal MS-AFP and AF-AChE	Liveborn; spinal surgery
Dabra *et al.*[13]	40	28	Routine	None	?	Liveborn; asymptomatic
Cherif *et al.*[6]	32	22	Routine	None*	?	Liveborn; asymptomatic
	30	22	Spina bifida	None	?	Liveborn; asymptomatic
Sonigo-Cohen *et al.*[7]	28	20	Routine	Meningocele	Normal AF-AFP and AF-AChE	Liveborn; spinal surgery
	29	22	Routine	Hypotrophic and hypomotile right foot*	Normal AF-AChE	Liveborn; physiotherapy
	38	22	Routine	Abnormal vertebral segmentation at the lumbosacral region with hemivertebra, single pelvic kidney*	Normal AF-AChE	TOP at 31 weeks; severe anomalies of vertebral segmentation in the lumbar region, single pelvic kidney, anal atresia, unicornuate uterus
Biri *et al.*[14]	34	15	Routine	Scoliosis	Elevated MS-AFP	TOP

*Fetal magnetic resonance imaging was also used. AF-AChE, amniotic fluid acetylcholinesterase; AF-AFP, amniotic fluid alpha-fetoprotein; GA, gestational age; MA, maternal age; MS-AFP, maternal serum alpha-fetoprotein; TOP, termination of pregnancy.

malformations such as spina bifida, hemivertebra, butterfly vertebra or kyphoscoliosis. The pathology was first described by Cruvelhier in 1853 (see Caspi *et al.*[2]). Two types of diastematomyelia have been defined: Type 1 diastematomyelia, where each hemicord has its own dural sheath, and Type 2 diastematomyelia, where both hemicords are covered by a common dural sheath[3]. Most of the lesions are located at the lower thoracic and the upper lumbar regions[1]. Sonographic features of diastematomyelia include widening of the spinal canal in the coronal plane, an additional echogenic focus traversing the spinal canal between the anterior and posterior walls in the axial plane and intact skin and soft tissues overlying the affected spinal segment[4]. Additionally, echogenic foci in the posterior aspect of the vertebral column are highly specific for diastematomyelia[5]. The prevalence of diastematomyelia is not known and cannot be calculated from our report because the majority of patients were referred to our institution for diagnosed spinal abnormalities.

On diagnosis of diastematomyelia in the fetus, the most pressing issue in terms of prognosis is to determine the presence of an associated spinal abnormality. Nine of the previously reported 26 cases of diastematomyelia had associated spinal abnormalities (Table 2). Sepulveda *et al.*[4] stated that a normal appearance of spinal curvature, intact skin overlying the spine and the absence of cranial signs of open spina bifida such as the banana and lemon signs, are reliable markers for the diagnosis of isolated diastematomyelia. They also stated that normal maternal serum α-fetoprotein level is an indicator of the integrity of the spinal canal[4].

We used the biochemical markers AF-AChE and AF-AFP to determine whether diastometamyelia was the only abnormality present in the fetus. AF-AChE is a reliable marker for the exclusion of open spina bifida. Fetal MRI is another diagnostic tool for determining associated spinal abnormalities. Three patients presented in the previous reports were also evaluated with fetal MRI, however, this did not change the diagnosis or add any information to the initial sonographic findings[6,7].

The cases reported by Pachi *et al.*[8] and Boulot *et al.*[9], two cases reported by Allen and Silverman[3], the first two cases reported by Sonigo-Cohen *et al.*[7] and the eight cases in our report were analyzed for chromosomal abnormalities. None of these 14 cases had chromosomal abnormalities.

Three women among the previously reported cases had obstetric complications in their pregnancy: maternal diabetes, maternal use of acyclovir and triplet pregnancy. Although the etiology of this rare abnormality is not known, it is difficult to assess the impact of these problems as potential causes of diastematomyelia[1,10,8].

The mean gestational age at diagnosis for previously reported cases of diastematomyelia was 20.9 ± 4.3 weeks (Table 2). The mean gestational age at diagnosis in our report was similar, at 21.1 ± 3.9 weeks. Although diastematomyelia is usually diagnosed during second-trimester scanning, one of the cases in our report (Case 3) was diagnosed during the 13th week of gestation. To our knowledge this is the earliest diagnosis of diastematomyelia and the first diagnosis made during first-trimester ultrasound screening.

CONCLUSIONS

Diastematomyelia is a rare abnormality of the spinal canal which may be associated with other spinal anomalies. Six of the eight cases from our report and 12 from the 26 previously reported cases had normal biochemistry and/or no other additional abnormality. These 18 fetuses had a favorable outcome, from which we conclude that isolated cases of diastematomyelia have a favorable prognosis. Diastematomyelia can be suspected sonographically as early as the first trimester. If diastematomyelia is found, amniocentesis for AF-AChE and AF-AFP is a reliable indicator of spinal integrity. Intrauterine recognition of diastematomyelia should facilitate appropriate management of the disease, which is important for the prevention of neurological sequelae.

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