

# Prenatal Diagnosis of Diastematomyelia Associated with Dandy-Walker Malformation: Case Report

## Dandy-Walker Malformasyonu ile Birliktelik Gösteren Diastematomiyeli Olgusunun Prenatal Tanısı

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**ABSTRACT** Diastematomyelia is a rare malformation characterized by division of the spinal cord into two columns by a cartilaginous or osseous spur. Even though anomalies of the vertebral column have been reported with diastematomyelia, a case associated with Dandy-Walker malformation has not been reported before in the literature review. In this report, we describe a case of diastematomyelia associated with Dandy-Walker malformation. Ultrasonographic examination revealed that fetal spine has abnormal widening of upper lumbar part on coronal scan with a central echogenic focus at area of widening and on transverse scan no posterior defect or soft tissue mass. Fetal magnetic resonance imaging examination detected the same findings as the ultrasonographic scan. Karyotype analyses were normal. The parents were counselled and elected to termination of pregnancy. At 22 weeks, she was delivered a 330 g female fetus. After delivery, spinal radiography and the fetal autopsy confirmed the diagnosis of diastematomyelia associated with Dandy-Walker malformation.

**Key Words:** Neural tube defects; Dandy-Walker syndrome; ultrasonography

**ÖZET** Diastematomiyeli, spinal kordonun kırıkdaksı ya da kemiksi bir çıkıntı ile iki kolona ayrılması ile karakterize nadir bir malformasyondur. Her ne kadar diastematomiyeli ile birlikte vertebral kolon anomalileri bildirilmiş olsa da, Dandy-Walker malformasyonu ile ilişkili bir olgu literatür derlemesinde daha öncesinde bildirilmemiştir. Bu olgu sunumunda, Dandy-Walker malformasyonu ile ilişkili bir diastematomiyeli olgusu tarif edilmiştir. Ultrasonografik incelemede, koronal kesitte fetal spinada üst lumbar bölgede anormal genişleme ile birlikte genişleme alanında santral bir ekojenik alan saptanmış ve transvers kesitte posteriorda defekt ya da yumuşak doku kitlesi belirlenmemiştir. Fetal magnetik rezonans incelemede ultrasonografik incelemedeki aynı bulgular saptanmıştır. Karyotip verileri normaldir. Aileye bilgi verilmiş olup, aile gebelik sonlandırılmasını tercih etmiştir. 22. gebelik haftasında 330 g, kız bebek doğurtulmuştur. Doğum sonrasında spinal radyografiler ve fetal otopside Dandy-Walker malformasyonu ile birliktelik gösteren diastematomiyeli tanısı doğrulanmıştır.

**Anahtar Kelimeler:** Nöral tüp defektleri; Dandy-Walker sendromu; ultrasonografi

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**D** iastematomyelia is a rare malformation characterized by division of the spinal cord into two columns by a cartilaginous or osseous spur.<sup>1</sup> 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. The malformation may be isolated or associated with other seg-

mental anomalies of the vertebral bodies or visceral malformations (horseshoe or ectopic kidney, utero-ovarian malformation, and anorectal malformation).<sup>2</sup>

Prenatal diagnosis is possible by ultrasound and magnetic resonance imaging (MRI). The prenatal diagnosis by ultrasound has first been introduced by Williams et al in 1985.<sup>3</sup> Sonographic features of diastematomyelia include widening of the spinal canal in the coronal plane, an additional echogenic focus transversing the spinal canal between the anterior and posterior walls in the axial plane and intact skin and soft tissues overlying the affected spinal segment.<sup>3,4</sup>

Dandy-Walker malformation (DWM) is described as a triad of cystic dilation of the fourth ventricle, complete or partial agenesis of the cerebellar vermis, and an enlarged posterior fossa with elevated tentorium.<sup>5</sup> The incidence of DWM is thought to be 1:25.000-30.000.<sup>6</sup> The syndrome usually results from a developmental failure of the the roof of the 4<sup>th</sup> ventricle during embryogenesis. Approximately 90 % of patients have hydrocephalus, and a significant number of children have associated anomalies including agenesis of the posterior cerebellar vermis and corpus callosum.<sup>7</sup>

In this report, we describe a case of diastematomyelia associated with Dandy-Walker malformation in the 21st gestational week using routine ultrasound scanning supplemented by fetal MRI.

## CASE REPORT

A 19 year-old gravida 3, para 2 pregnant woman was referred to our perinatology unit at 21 weeks 4 day of gestation for an abnormal obstetric ultrasound. She had a previous healthy child born with a cesarean section and an abortion at 8 weeks of gestation last year. Both 2-dimensional (2-D) and 3-dimensional (3-D) ultrasonographic examinations were performed with Sonoace X8, Samsung Medison SA Co. Seoul, South Korea, 2-6 MHz. 2D and 3D ultrasonographic assessments revealed that fetal spine had abnormal widening of upper lumbar part on coronal scan with a central echogenic focus at area of widening and on transverse scan no pos-

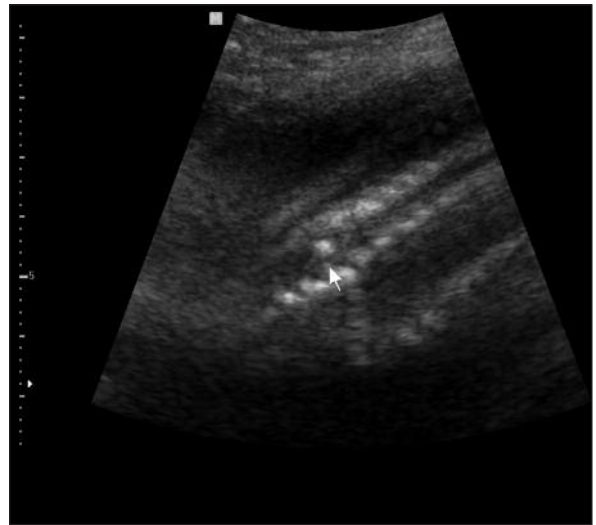
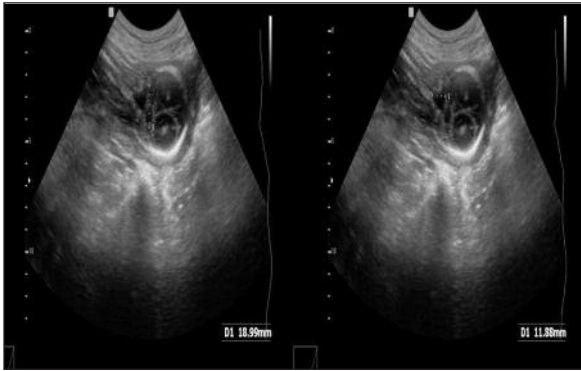


FIGURE 1: Ultrasound images at 21 weeks' gestation showing an extra echogenic focus in coronal section (arrow).

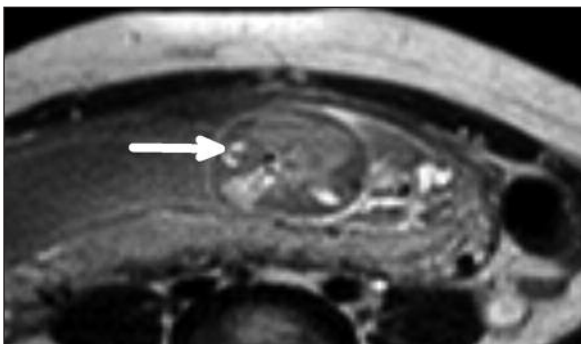


FIGURE 2: 3D image showing splitting of spinal cord.

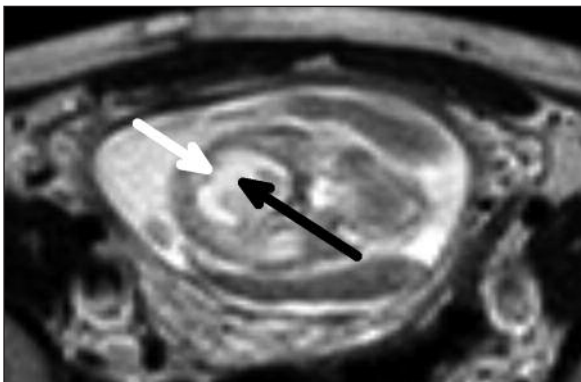
terior defect or soft tissue mass (Figures 1, and 2). Ventriculomegaly (lateral ventricle 13.2 mm), enlarged cisterna magna (14 mm) and cerebellar vermician hypoplasia were also diagnosed during the ultrasonographic investigation (Figure 3). Axial T2 weighted MR image of the fetal spine at lumbar level demonstrated diastematomyelia and image of the fetal posterior fossa showed mega cisterna magna and inferior vermician hypoplasia (Figures 4, and 5). Karyotype analyses, levels of amniotic fluid alpha-fetoprotein (AF-AFP) and acetylcholinesterase (AF-AChE) were normal. The parents were counselled by a multidisciplinary team regarding the implication of diastematomyelia with Dandy-Walker malformation and elected to termination



**FIGURE 3:** Image showing enlarged cisterna magna and cerebellar verian hypoplasia.



**FIGURE 4:** Axial T2 weighted MR image of the fetal spine at lumbar level demonstrates diastematomyelia. The spinal canal is divided by osseus septa (arrow).



**FIGURE 5:** Axial T2 weighted image of the fetal posterior fossa shows mega cistern magna (white arrow) and inferior verian hypoplasia (black arrow).

of pregnancy. At 22 weeks, she was delivered a 330 g female fetus. After delivery, spinal radiography and the fetal autopsy confirmed the diagnosis of diastematomyelia associated with Dandy-Walker malformation.

## DISCUSSION

Diastematomyelia is a rare abnormality of the neural tube and is usually associated with other spinal malformations.<sup>8-11</sup> Lesions are usually located on the lower thoracic and upper lumbar parts of the spine. Besides, diastematomyelia in the cervical spine has been reported before.<sup>12</sup>

Even though anomalies of the vertebral column such as spina bifida, kyphoscoliosis, butterfly vertebra, and hemivertebra have been reported with diastematomyelia, a case associated with Dandy-Walker malformation has not been reported before in the literature review.<sup>13</sup> The importance of our case is the diagnosis of diastematomyelia and Dandy-Walker malformation in the same case using routine ultrasound.

Identification of an extra echogenic posterior focus in the spinal canal in the axial plane and widening of the spinal canal in the coronal plane are specific prenatal signs of diastematomyelia. Sepulveda et al. 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 banana and lemon signs, are reliable markers for the diagnosis of isolated diastematomyelia.<sup>14</sup> They also stated that normal maternal serum  $\alpha$ -fetoprotein level is an indicator of the integrity of the spinal canal. In our case report, levels of amniotic fluid  $\alpha$ -fetoprotein (AF-AFP) and acetylcholinesterase (AF-AChE) were evaluated. The levels were normal enough to exclude the diagnosis of open spina bifida.

Advanced imaging techniques such as CT and MRI can provide detailed information about spinal cord anatomy. Breningstall et al have performed MRI in 45 patients with the diagnosis of myelomeningocele and diagnosed two cases of diastematomyelia.<sup>15</sup> In our case, we performed MRI to confirm our postnatal diagnosis. But addition of MRI as a diagnostic tool neither changed the initial diagnosis nor added any further information.

There are several abnormalities associated with Dandy-Walker malformation.<sup>16,17</sup> The most common are ventriculomegaly (70-90%) and car-

diac defects. Midline defects such as agenesis of corpus callosum (20-25%), holoprosencephaly (25%), dysplasia of the cingulate gyrus (25%), encephalocele (7%), syringomyelia, Klippel-Feil deformity, spina bifida, polycystic kidney disease and facial dysmorphism. However, as to our knowledge, no case of Dandy-Walker associated with diastematomyelia has been reported before. In this case, the concomitant presence of these two malformations has a special impact on prenatal diagnosis. DW malformation is described as a triad of cystic dilation of the fourth ventricle, complete or partial agenesis of the cerebellar vermis, and an enlarged posterior fossa with elevated tentorium and fetal diastematomyelia is described as a form of spinal dysraphism characterized by a complete or incomplete division of the spinal cord by an osseous or fibrocartilaginous septum. For this reason, an increased pressure of the intracranial structures may result in complete or incomplete division of the

spinal cord. In such situations, the accurate diagnosis of antenatal conditions is essential not only for management of the current pregnancy but also for future pregnancies to aid in the identification of the inheritable diseases and provide information for accurate pre-natal counselling. Besides we had not much knowledge since such a combination has not been reported before, so we were not able to counsel the patient in detail.

Diastematomyelia and Dandy-Walker malformation concurrency is unique. It seems that diastematomyelia without other detectable ultrasonographic markers carries a good prognosis, but in cases associated with other rare abnormalities, more case series are needed to counsel the patients more accurately. Also the parents should be counselled by a multidisciplinary team and be aware of the neurological and orthopedic involvement occasionally seen in these cases.

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