

편측척추의 산전진단: 3차원 초음파의 역할

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Prenatal Diagnosis of Hemivertebra: Role of Three-Dimensional Ultrasonography

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Hemivertebrae is the most common form of congenital scoliosis, although the overall prevalence is rare. We report a 28-year-old primigravida who sought evaluation at our institution for spinal deformity at 21 weeks gestation. Using two- and three-dimensional ultrasonography, an abnormal shaped thoracic vertebra with a missing rib, deviated alignment, and scoliosis was demonstrated in the fetus. After delivery due to premature rupture of membranes, an infantogram confirmed a single hemivertebra at T11, which was in agreement with the prenatal sonographic findings. Here we discuss the value of three-dimensional ultrasonography as an essential diagnostic tool for the prenatal diagnosis of a hemivertebra.

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The spine is a major structural support of the body and functions as a safe passage of the central nervous system to the periphery. For proper formation of the spine, there are complex events occurring concurrently and serially during the embryonic period. Therefore, if there is a break in normal development or any insult that disrupts the formation of the spine, congenital spine deformities will occur. Spine deformities are classified into three types (neural tube defects, defects of segmentation, and defects of formation).¹ The defects of formation involve missing structural parts in the vertebrae that lead to hemivertebrae.

Hemivertebra, part of a formation defect of the spine, is one of the common causes of congenital scoliosis, albeit a rare abnormality. We managed a gravida at 21 weeks gestation who

was diagnosed by ultrasonography to have a fetus with a single hemivertebra. We therefore present the sonographic and radiologic features of this abnormal condition. We will also discuss the values of conventional and three-dimensional (3D) ultrasonography as essential diagnostic tools for the prenatal diagnosis of a hemivertebra.

Case Report

A 28-year-old primigravida was referred to our tertiary center for evaluation of a suspected spine anomaly at 21⁺³ weeks gestation. The patient and her spouse had no medical, family, infectious, or drug or toxin exposure histories. Two-dimensional (2D) and 3D ultrasonography with a Voluson 730 Expert (GE Medical Systems, Milwaukee, WI, USA) revealed a single fetus. The fetal biometry, biparietal diameter, head and abdominal circumferences, and femur length were compatible with 20⁺⁶

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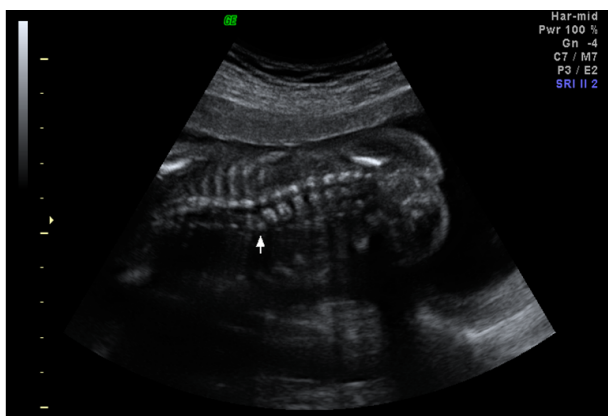


Fig. 1. Sonographic coronal view of the spine with thoracic hemivertebrae (arrow).

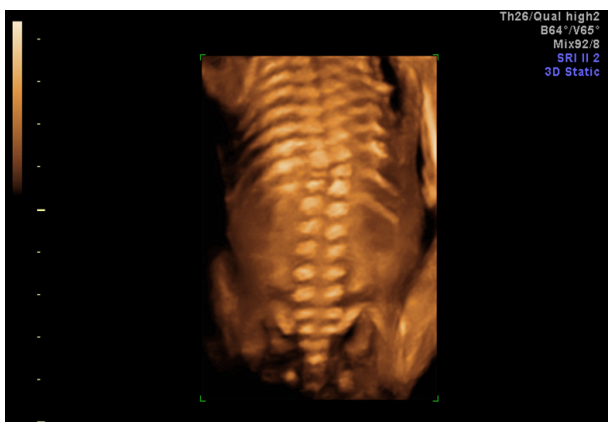


Fig. 2. Three dimensional ultrasound showing thoracic hemivertebra with one missing rib.

weeks gestation. The placenta was located posteriorly and the amniotic fluid volume was appropriate for gestational age. The fetal brain, face, heart, stomach, intestines, kidneys, bladder, extremities, and genital structures appeared normal. In the coronal and sagittal planes, one triangular-shaped smaller vertebral body was visible forming a distorted parallel line of the lower segment of the spine (Fig. 1). Accompanying 3D ultrasound, a slightly deviated alignment of the spine forming a convexity on the right side was demonstrated on the coronal view. There was one wedge vertebra between two relatively normal vertebrae in the thoracic region and one rib was missing on the opposite side of the vertebra (Fig. 2). Comparing 2- and 3D ultrasonography, the shape and margin of each vertebra and the missing rib was more clearly defined on 3D than 2D ultrasonography. We suspected



Fig. 3. Fully segmented 11th thoracic hemivertebra by infantogram.

an 11th thoracic hemivertebra, scoliosis, and a missing rib. One week later, due to premature rupture of membranes, the parents opted for a termination of the pregnancy. Prostaglandin agents were administered orally and a male fetus weighing 400 grams was delivered the following day. The gross examination showed no other fetal abnormalities with the exception of a lateral curvature of the spine and scoliosis. On an infantogram, a single, fully segmented, hemivertebra was observed at the 11th thoracic segment of the spine and one missing rib, in agreement with prenatal sonographic findings (Fig. 3). The parents declined fetal karyotyping and autopsy.

Discussion

Hemivertebrae are the most common cause of congenital scoliosis, with a prevalence of 5-10 in 10000 live births, with a female preponderance.² The etiology of hemivertebrae is not established, but maternal exposure to toxins, such as carbon monoxide, maternal diabetes, and ingestion of certain drugs as anti-epileptic drugs have been postulated as possible causes.³⁻⁵ There are several reports of a correlation with genetic inheritance, such as concordant hemivertebrae in monozygotic twins, chromosomal deletions, and translocations.^{2,6-7} Greater than two-thirds

of those with hemivertebrae have associated anomalies, such as VACTERL, Jarcho-Levin, Klippel-Feil syndromes, and Sprengel deformities.⁸⁻¹⁰ Abnormalities of other organ systems often occur at the same time, most frequently renal anomalies followed by cardiac anomalies because of the common mesodermal origin, including the skeletal system.¹

There are four different types of hemivertebrae (fully segmented, semi-segmented, non-segmented, and incarcerated).¹ This case was categorized as fully segmented, which means a defected vertebra without fusion of the vertebrae above or below. According to McMaster et al.,¹¹ greater than one-half are the fully segmented type which results in progressive curvature if there is no proper treatment; the other three types infrequently require treatment.¹²

A single hemivertebra, as in this case, has a more favorable outcome than a hemivertebra with multiple anomalies or an associated syndrome. The prognosis of a newborn with a hemivertebra depends on the location, type, and rate of deterioration after birth. The natural history of congenital scoliosis has been reported to be non-progressive in 25% of case, while the remainder of the cases are mild or highly progressive and eventually cause neurologic impairment or cardiopulmonary compromise.¹² Therefore, earlier detection and treatment of hemivertebrae is important for better outcomes.

Prenatal diagnosis of congenital spine deformities requires a thorough sonographic examination with sagittal, coronal, and transverse views. The prenatal diagnosis of hemivertebrae is generally made at 15-22 weeks gestation, as in the current case.⁹¹³ However, because ossification occurs as early as the 12th week of gestation, it might be possible to detect the defect at this point and the earliest reported case with a single hemivertebra has been the 13th week of gestation.¹⁴ In the current case, spatial reconstruction 3D ultrasound in the surface mode provided a detailed picture of the vertebral anatomy at the precise point of the hemivertebra and missing rib. Spatial reconstruction with surface mode is based on a volume rendering method and gives examiners clear anatomic relationships which are unavailable using 2D ultrasound. The reason for not using the skeletal mode of fetal bone is incomplete bone formation at this gestational age. Thus,

bone mineralization is not complete until fetuses are at term or when the fetus is born. Visualization of abnormalities by the skeletal mode has limitations, thereby resulting in differences in the shape of hemivertebrae between 2D and the surface mode of 3D ultrasound. As a result, we were able to confirm the shape of the vertebral body and missing rib that were not clearly visible on gray scale images. Therefore, in the case of suspected fetal skeletal abnormalities based on 2D scanning, the surface mode of 3D images is an essential step that is recommended for precise prenatal diagnosis.

In conclusion, based on our experience with a single hemivertebra, 3D ultrasonography is a detailed and valuable tool for the accurate diagnosis of suspected vertebral abnormalities.

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국문요약

편측척추는 매우 드물지만 선천성 척추측만증의 가장 흔한 원인중 하나이다. 임신 21주에 척추 기형이 의심되어 전원된 28세 초산모의 2차원 초음파 검사를 통해 편측 흉추를 확인, surface mode의 3차원 초음파검사에서는 11번 흉추의 편측 척추, 환측 늑골형성 부전 및 척추측만증을 산전진단하고 출생후 방사선 검사로 확인하였기에 문헌고찰과 보고하는 바이다.

중심 단어 : 선천성 편측척추, 산전진단, 3차원 초음파검사
