Prenatal Diagnosis of a Unilateral Hypoplastic Vertebral Arch by 2- and 3-Dimensional Sonography

A 25-year-old woman, gravida 1, para 0, was referred to our hospital at 24 weeks’ gestation for routine second-trimester sonography. The patient had no notable medical history, no history of exposure to any toxic substances, and no notable family history. The couple was nonconsanguineous.

Sonography at 12 weeks’ gestation revealed no abnormality. Second-trimester serum marker values were normal.

At 24 weeks, sonography using a multifrequency transabdominal volume transducer (Voluson E8; GE Healthcare, Zipf, Austria) revealed a live fetus. A mid-sagittal scan of the spine showed improperly aligned vertebrae in the lumbosacral part. On a transverse section, only the right half of the L4 vertebral body was present, raising suspicion of a L4 hemivertebra (Figure 1A and Video 1), but the bilateral vertebral arches of L4 were symmetric and normal in size. On a coronal view of the spine,

Figure 1. Unilateral hypoplastic vertebral arch. A and B. Two-dimensional images of the fetal spine at 24 weeks’ gestation. A. Transverse section of L4 showing a hemivertebra (arrow). B. On a coronal view, an echogenic focus is wedged on the right side (arrow). C. Postmortem computed tomographic reconstruction showing the L4 hemivertebra (arrow) and hypoplastic L1 vertebral arch (arrowhead). D. Prenatal 3-dimensional rendering showing the smaller left L1 vertebral arch (arrow).
an irregular arrangement of the posterior spinal echogenic foci was noted from L1 to L3, with a decreased interechogenic space and a small echogenic focus wedging on the right side (Figure 1B). Three-dimensional (3D) volumes of the spine were acquired and saved with the volume box, including the thoracic and lumbar parts of the spine, at a volume angle of 65° with a standard preset image resolution. No other malformations could be detected. Thus, a diagnosis of multiple congenital vertebral malformations was made. Prenatal magnetic resonance imaging (1.5 T; GE Healthcare) showed no other anomalies, including intracranial structures, and provided no further information about the vertebrae. Amniocentesis was performed.

Because of the above-described congenital anomalies, the couple elected to terminate the pregnancy. A male fetus with no obvious external abnormalities was delivered at 25 weeks’ gestation. Postmortem computed tomography (CT) with 3D reconstruction showed the slightly misaligned lumbosacral part of the spine as well as the L4 hemivertebra and a hypoplastic L1 vertebral arch (Figure 1C). At the L4 level, the left half of the vertebral body was absent, whereas bilateral vertebral arches were normal. At the L1 level, the vertebral body was intact, but most of the left arch was missing, with only a small posterior remnant. Postmortem sonography was also done, showing that the left vertebral arch of L1 was a little smaller than the right side on a transverse section. These abnormal findings, especially the hypoplastic L1 vertebral arch, prompted us to reanalyze the stored prenatal 3D volumes using 4D View version 7.0 data management software (Luminary; GE Healthcare). On the rendered images in the transparent maximum mode mixed with the surface texture mode (70%/30%), bony structures were easy to recognize. The vertebrae were examined one by one, and the hypoplastic left L1 spinal arch could also be identified and was smaller than the opposite side (Figure 1D).

Microscopic examination demonstrated the L4 hemivertebra, with no ossification center on the left side. The left vertebral arch of L1 was hypoplastic with an ossification center smaller than that on the right side. The cartilaginous component of the L4 and L1 vertebrae were symmetric in size bilaterally. The spinal cord was not compressed at the level of the malformation, and the conus medullaris ended at the level of L3-L4 space.

A hemivertebra is one of the most common vertebral bony anomalies, resulting from failure of a vertebra to form on one side. It is not difficult to detect when multiple vertebrae are involved or accompanied by neural tube defects. Most cases reported in the literature typically showed that half of the vertebra, including both the vertebral body and arches, was missing. However, in our case, the bilateral arches of L4 were intact, whereas only half of its vertebral body was absent, which to our knowledge has not been reported in the prenatal sonographic literature. In the absence of other malformations, the prognosis of hemivertebra is principally based on vertebral continuity and stability, which are ensured by the vertebral arches. Therefore, differentiating a hemivertebra with bilateral intact arches from that with only a unilateral arch by 2-dimensional (2D) or 3D sonography may help predict the prognosis.

Hypoplasia of the unilateral vertebral arch without any defect of the vertebral body is an uncommon anomaly, which to our knowledge has not been reported in the literature. During embryologic development, there are 3 ossification centers involved in the formation of a normal vertebra. The incompletely ossified posterior centers may lead to hypoplastic vertebral arches. In our case, although the irregular arrangement of the posterior spinal echogenic foci was noted on prenatal sonography, the definite diagnosis was not obtained until the postmortem CT scan was performed. Postmortem sonography revealed that the asymmetry of the L1 arches was not quite as prominent in the axial plane, which may have been the reason why the lesion of the L1 arch was not detected by prenatal 2D sonography. Retrospective analysis of the 3D sonographic volumes showed that the left arch of L1 was obviously smaller on the rendered image, similar to the CT findings. Therefore, we believe that 3D sonographic volumes of the fetal spine should be obtained, and each vertebra should be carefully examined on rendered images because 3D sonography may provide more valuable information than 2D sonography in detecting subtle lesions of fetal vertebrae, as our case showed.

Although prenatal CT may provide some reassuring information in a fetus suspected to have congenital vertebral anomalies, its application in fetuses is restricted by radiation safety concerns. Fetal magnetic resonance imaging is informative in excluding central nervous system malformations, but it has not been proven to exceed dedicated 2D or 3D sonography for the detection of subtle vertebral anomalies, especially at an early gestational age, as shown in our case.

The irregular arrangement of the posterior vertebral ossification centers on coronal and sagittal views should be differentiated from diastematomyelia, which has similar sonographic findings. Although definite diagnosis can be difficult, the transverse sonographic view can be very helpful in the differential diagnosis because a vertebra in diastematomyelia has 3 posterior echogenic foci, with the central one protruding toward the skin or the neural canal.
In summary, we have reported a rare case of a unilateral hypoplastic vertebral arch and a hemivertebra on separate fetal lumbar vertebrae at 24 weeks’ gestation. To the best of our knowledge, a case of a single unilateral hypoplastic vertebral arch that was suspected on prenatal sonography and confirmed by postmortem CT and pathologic findings has not been reported previously. The misalignment of the echogenic foci of the spine detected by prenatal 2D sonography is not only a sign of a vertebral body defect such as a hemivertebra but also a manifestation of vertebral arch dysplasia, which has been rarely recognized before. Prenatal 3D sonography is essential in the localization and diagnosis of subtle lesions of the fetal spine and may provide prognostic information. For suspected cases, we recommend that 3D sonographic volumes of the fetal spine be acquired and each vertebra be carefully assessed because 3D sonography is useful in detecting subtle anomalies of the fetal spine that are not accessible on 2D sonography, as shown in this case.

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References