Prenatal assignation of lesion levels in neural tube defects by using ultrasonography

Case report and review of the literature

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✓Myelomeningoceles are routinely diagnosed prenatally by using ultrasonography. Because the level of the neural defect has been shown to correlate with functional and cognitive outcome in infants with myelomeningocele, it is of prognostic importance to identify the level prenatally. The authors report on a case in which the postnatal vertebral level of the myelomeningocele was more rostral than expected based on prenatal imaging.

KEY WORDS • myelomeningocele • spine bifida • fetus • spine • ultrasonography

ULTRASONOGRAPHY is commonly used in the in utero diagnosis of neural tube defects. We report on the use of prenatal ultrasonography to determine the spinal level of the neural defect in the fetus of a 22-year-old woman.

We also review the literature on the use of prenatal ultrasonography and discuss its accuracy compared with other imaging modalities.

Case Report

This 22-year-old Hispanic woman (Gravida 1, Para 0) was referred from another institution for an in utero neural tube defect. She underwent ultrasonography at 20 weeks’ gestation. The mother had been taking valproic acid for a seizure disorder throughout the pregnancy. Fetal cranial ultrasonography findings (Sonoline Elegra; Siemens, Issaquah, WA) included effacement of the cisterna magna, mild head molding, and a large cystic myelomeningocele. The prenatal anatomical level accurately predicted the neuromotor level when the child reached 1 year and at 3 to 4 years of age, with a lower correlation at 6 years of age.

The baby was delivered by elective cesarean section at 38 weeks. Physical examination revealed a full fontanel, incontinent of bowel and bladder incontinence, in addition to variable lower-extremity involvement. She elected to continue the pregnancy.

The baby was delivered by elective cesarean section at 38 weeks. Physical examination revealed a full fontanel, mild head molding, and a large cystic myelomeningocele saccus measuring approximately $6 \times 5 \times 4$ cm (Fig. 4), in the center of the placental area. The child had a motor functional level of paraplegia at L1–3 (quadriplegs function) clinically with more function on the left than right side. There was no pelvic tone, and the child seemed to be incontinent. On x-ray films, the L-5 VB was intact. There was bilateral talipes equino varus deformity was also noted. The child was not able to sit unassisted.
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**Discussion**

Neural tube defects are a common birth defect, despite increasing periconceptional use of folate, and are associated with variable degrees of disability in affected infants. The introduction of maternal α-fetoprotein and ultrasonography screening programs have led to increased prenatal detection and decreased birth incidence, to approximately 1 to 1.6 per 1000 births.2,3 Open spina bifida has a variable prognosis, and prenatal detection by ultrasonography raises prognostic and counseling issues. Important postnatal prognostic variables include the level and type of lesion; presence of ventriculomegaly; associated anomalies, such as karyotype abnormalities; and type of surgical intervention. As fetal surgical procedures become more commonplace, prenatal identification of features is likely to become more important.2 Identification of these factors prenatally aids in parental counseling and decision making.

Ultrasonography evaluation of the fetal spine requires identification of the three vertebral ossification centers at each vertebral level, with the posterior ossification centers forming the neural arches normally either converging or parallel to each other in the transverse plane. Imaging of the fetal spine is ideally performed in three planes: axial, coronal, and sagittal.5 The axial images are believed to be the most sensitive for detection of spina bifida because all three ossification centers and skin integrity may be evaluated simultaneously.5 Spina bifida may be detected ultrasonographically in this plane as the divergence of the posterior ossification centers forming the fetal neural arch and in the coronal plane as the widening of the usually parallel ossification centers. Helpful associated features include the presence of a cystic meningeal sac and characteristic cranial abnormalities of the Chiari Type II malformation: reduced cerebellum, effacement of the cisterna magna, and ventriculomegaly.

It is accepted that the postnatal anatomical level has ramifications for neuromotor, functional motor, and cognitive outcome in affected neonates. Authors of recent reports have also shown a correlation with prenatal lesion level, which therefore has important implications in parental counseling regarding prognosis. It is clinically important to identify the affected vertebral level antenatally.2,3,5-7

Anatomical level can be assigned in the second trimester by using ultrasonography and counting cephalad from the VB at the level of the iliac wings, assumed to be the S-1 VB in most cases and confirmed by counting caudally from the T-12 VB, adjacent to the 12th rib (assuming 12 thoracic ribs).4-9,12 Kollias, et al.11 examined 28 infants with prenatally detected myelomeningocele and found a correlation between ultrasonography demonstrated and pathological levels in 64% and within one spinal level in 79%. They concluded that ultrasonography can "in most cases, allow accurate prediction of the level of the spina bifida lesion and the severity of neuromotor handicap." Recent data suggest that there may be variable accuracy in identification of anatomical level; Woods, et al.16 demonstrated poor exact agreement between prenatal ultrasonography results and postnatal radiographs (32.9%), but good correlation to within one level (78.5%), with a tendency to overestimate the myelomeningocele level based on ultrasonography findings. Despite modification of the ultrasonographic technique, accuracy was not improved and they concluded that a different modality may be required to improve accuracy, particularly with increasing consideration of fetal surgical techniques. Coniglio, et al.7 evaluated functional motor outcome with respect to prenatal anatomical level of myelomeningocele. The prenatal anatomical level accurately predicted the neuromotor level when the child reached 1 year and at 3 to 4 years of age, with a lower correlation at birth attributed to difficulty in assessing neuromotor status in neonates. Prenatal anatomical level was also found to predict accurately functional motor outcome at 3 to 4 years. They concluded that lesion level determined prenatally allowed accurate prediction of neuromotor level, radiographic level, and functional motor outcome in affected children, but cautioned that other factors have a role in determining independent ambulation.7 Biggio, et al.2 assessed ambula-

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**Fig. 1.** Axial ultrasonography study obtained at the level of the iliac crests (S-1), demonstrating a septated cystic myelomeningocele sac and splaying of the posterior elements of the spine.

**Fig. 2.** Axial ultrasonography study obtained at L-5, demonstrating an intact skin line and normally converging posterior ossification centers.
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The 12th rib is identified (arrow).

Fig. 3. Coronal ultrasonography study obtained of the lumbosacral spine, demonstrating splaying of ossification centers of the sacral spine and the cystic myelomeningocele sac; it is difficult to rule out definitely lower lumbar spine involvement. The 12th rib is identified (arrow).

In a subsequent paper, Coniglio, et al. determined that cognitive developmental outcomes correlated significantly with degree of ventriculomegaly. Increasing severity of ventriculomegaly was associated with lower cognitive developmental quotients in children with prenatally diagnosed myelomeningocele. Additionally, they found a significant correlation between the prenatally determined level of dysraphism and cognitive developmental testing, with lower level lesions being associated in most cases with better outcome.

It can be seen that there is variation among reports by authors with respect to accuracy in assignment of lesion level, which is likely due in large part to the inherent operator dependence of ultrasonography as well as the often difficult anatomy. There are also discrepancies between postnatal surgically assigned level and radiographic level, with the surgical defect often appearing smaller than the radiographically identified abnormality. These results are of considerable interest because lesion level has been shown to influence prognosis in neuromotor, functional motor, and cognitive outcomes in these infants and therefore prenatal counseling may be unintentionally misleading. Additionally, the study comparing the postnatally assigned radiographic level with functional status demonstrated incomplete agreement between these assessment modalities, raising further issues regarding counseling.

Authors of recent studies have attempted to determine whether accuracy can be improved by 3D techniques. Three-dimensional ultrasonography has been shown to provide additional information, improved accuracy in the assignment of level of defect, and occasionally demonstration of anomalies not seen on 2D imaging. Early experience with 3D imaging suggested that use of three orthogonal planes of imaging, were they demonstrated the anatomical level of the lesion, allowed a direct comparison of ultrasonography with postnatal MR imaging in fetuses with spina bifida. The authors also demonstrated the utility of 3D imaging in the evaluation of fetal myelomeningocele and the cystic myelomeningocele sac; it is difficult to rule out definitely lower lumbar spine involvement.

Fig. 4. Postnatal photograph showing a large cystic myelomeningocele sac at the lumbosacral level.
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orthogonal planes facilitated diagnosis of the nature and anatomical level of the defect and that 3D reconstructions demonstrated the extent of the lesion better and occasion-
ally allowed a diagnosis not possible by either 2D or non-
reconstructed 3D ultrasonography.15 Advantages of 3D ultrasonography may be due to improved diagnostic confi-
dence in level assessment, ability to rotate volume data acquired using a transvaginal probe, and shortened duration of imaging time because of the ability to store the examination data and reconstruct the images later. Johnson, et al.16 reported that lesion level could be determined more accu-
rately with 3D ultrasonography in three of five cases of neural tube defects. The technical limitations of the 3D technique were emphasized, particularly artifact caused by fetal movement, and limited imaging results when the fetus is pressed against the uterine wall or placenta, or when there is a lack of amniotic fluid around the region of the inter-
est.13,14 Riccabona, et al.,14 found that 3D ultrasonography facilitated understanding of complex anatomy relating to site and extent of abnormality. In addition, the ability to change rendering modes allowed display of different struc-
tures within the same data set, which aided in diagnosis. Lee, et al.,15 evaluated the accuracy of 3D compared with 2D ultrasonography in determining the extent of neural tube defects; they used postnatal plain radiographic films or MR images for comparison. They report agreement to within one vertebral segment in six of nine infants with 2D and eight of nine infants with 3D imaging. Multiplanar views proved to be more useful than rendered views for lo-
calizing neural tube defects in this group. It is clear that the use of 3D ultrasonography for prenatal determination of lesion level in neural tube defect requires further evaluation but it is likely be a useful adjunct to routine 2D imaging. Aaronsen, et al.,11 studied prenatal ultrasonography with MR imaging in fetuses with spina bifida. They compared prenatal ultrasonography images with postnatal radiographs and discovered that the findings agreed within one spinal level in 79% (55 of 70) of cases and that when prenatal MR images were compared with postnatal radiographs, the findings agreed in 82% (31 of 38) of cases. Findings on postnatal MR images and those on postnatal spinal radiographs agreed within one spinal level in 100% (50 of 50, κ = 1) of cases.

Conclusions

It is prognostically important to assign the correct verte-
bral level in cases of neural tube defect detected prenatally because there are significant implications in functional and cognitive outcome in affected infants. It is, however, appar-
ent that prenatally assigned levels may vary by two to three segments from postnatal assigned levels, with significant differences in expected outcome, even in the hands of experi-
enced operators. Additionally, even with correct assign-
ation of the bone defect level, the postnatal functional sta-
tus may vary from radiographically assessed level, often being higher (worse).15 In the case presented here, the in-
fant was born with a functional level three to five VBs higher than expected based on the 20-week ultrasonogra-
phy study, which was probably related to tethering of the spinal cord. Knowledge of the likely outcome may influ-
ence the decision to continue a pregnancy. Medical care providers who are counseling parents of neonates with me-
ningomyelocele should be aware of inherent inaccuracies in prenatal assessment and that postnatal functional status may be worse than expected, even with accurate designa-
tion of the bone defect level.

References

1. Aaronsen OS, Hernandez-Shulman M, Bruner JP, Reed GW, Tulipan NB: Myelomeningocele: prenatal evaluation—com-
3. Bonilla-Musoles F, Machado LE, Osborne NG, Muñoz EA, Ra-
gas, Blanes J, et al: Two- and three-dimensional ultrasound in malformations of the medullary canal: report of four cases. Pre-
4. Budorick NE, Pretorius DH, Grafe MR, Lou KV: Ossifica-
5. Budorick NE, Pretorius DH, Nelson TR. Sonography of the fe-
tal spine: technique, imaging findings, and clinical implica-
6. Coniglio SJ, Anderson SM, Ferguson JE: Relationships be-
7. Coniglio SJ, Anderson SM, Ferguson JE: Functional motor out-
come in children with myelomeningocele: correlation with anat-
10. Johnson DD, Pretorius DH, Riccabona M, Budorick NE, Nel-
11. Kollias SS, Goldstein RB, Cogen PH, Filly RA: Prenatally de-
14. Riccabona M, Johnson D, Pretorius DH, Nelson TR: Three di-