Intracranial Translucency and Spinal Cord Defects: Early Prenatal Diagnosis of Diastematomyelia

A 33-year-old woman, gravida 2, para 1, presented at a gestational age of 13 weeks 2 days for a routine 11- to 13-week examination for assessment of the risk of chromosomal abnormalities. Her obstetric and medical histories were unremarkable. Her fetal biometric measurement (crown-rump length, 61 mm) and nuchal translucency (1.8 mm) were normal.

In the midsagittal view of the posterior brain, the typical 4 lines were present, but the borders of the intracranial translucency were not easily identifiable, mainly because the fetal head was predominantly in a hyperextended and rotated position (Figures 1 and 2A). The brain stem diameter (4 mm) and brain stem diameter/brain stem-to-occipital bone diameter ratio (1.4) were increased. Careful examination of the fetal spine showed widening of the vertebral bodies at the lumbar region and the presence of an echogenic area in the spinal canal, features highly suggestive of diastematomyelia (Figure 2B). The fetal karyotype was normal (46,XX).

The parents were extensively counseled and opted for termination of the pregnancy. Postmortem examination confirmed complete type 1 diastematomyelia with a cartilaginous diaphragm in the spinal cord and spina bifida occulta.

Diastematomyelia is a rare congenital spinal disorder characterized by a sagittal cleft in the spinal cord with splaying of the posterior vertebral elements, usually in the lower thoracic and upper lumbar regions of the spine.1 The complete or incomplete division of the spinal cord into two hemicords is caused by an osseous, cartilaginous, or fibrous septum in the central portion of the spinal canal.

Prenatal diagnosis of diastematomyelia has been reported mainly in the second trimester.2–6 The ultrasonographic features include widening of the posterior ossification centers of the vertebrae, the presence of an echogenic focus traversing the spinal canal, intact skin and soft tissues overlying the affected spinal segment, and compression and division of the spinal cord into two columns.5 Echogenic foci in the posterior aspect of the vertebral column are highly specific for diastematomyelia.6 Isolated diastematomyelia has intact posterior soft tissues in the transverse scanning plane and normal maternal serum α-fetoprotein levels. Associated abnormalities are meningomyelecele, Arnold-Chiari malformation, hydrocephalus, abnormal vertebral segmentation/hemivertebra, and renal abnormalities. The clinical features of isolated diastematomyelia include skin lesions (hypertrichosis, hemangiomas, dermal sinuses, and subcutaneous lipomas), neurologic symptoms (back pain, functional disorders of the gastrointestinal and urogenital systems, and wasting or weakness of the lower extremities), and orthopedic problems (scoliosis).

In the last few years, sonography of the posterior brain in the midsagittal view at 11 to 14 weeks has shown potential for early detection of open neural tube defects. Chaoui et al7 described the fourth ventricle and named it intracranial translucency. They observed that in cases with open neural tube defects, intracranial translucency becomes difficult to identify because of the caudal displacement of the brain stem.7 Another group of investigators reported that the brain stem diameter/brain stem-to-occipital bone diameter ratio (Figure 1) invariably increased to greater than 1.0 in cases with open neural tube defects.8 Papastefanou et al9 studied the posterior brain and found both measurements of intracranial translucency and the cisterna magna to be highly reproducible at 11 to 14 weeks. They described the normal appearance of the posterior brain as the presence of 4 almost parallel lines (the 4-line view; Figure 1). A prospective study on 1300 fetuses using the 4-line view of the posterior brain to screen for neural tube defects at 11 to 14 weeks showed that examination of the posterior brain is feasible during the routine first-trimester examination and reported 2 true-positive cases and the first false-negative case of neural tube defects.10

Figure 1. Normal appearance of the posterior brain in a midsagittal view. Note the presence of the 4 parallel lines (the 4-line view) consisting of the following structures: brain stem (distance between lines 1 and 2), intracranial translucency (distance between lines 2 and 3), cisterna magna (distance between lines 3 and 4), and brain stem-to-occipital bone diameter (distance between lines 2 and 4).
Posterior brain abnormalities in the first trimester have not been previously reported in association with diastematomyelia. In fetuses with open neural tube defects, early herniation of the brain stem is the presumed pathophysiologic mechanism underlying the changes in the posterior brain, and this condition may have been true in our case as well.

This report adds to the emerging evidence that the midsagittal view of the posterior brain will be an important tool in the early diagnosis of neural tube defects and possibly other anomalies of the brain and spine.

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