

Fetal diagnosis of spondylocostal dysplasia: Limits of conventional fetal ultrasound & MRI in diagnosing anomalies

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We present a case of postnatally recognized spondylocostal dysplasia that was prenatally misdiagnosed as fetal thoracolumbar kyphoscoliosis secondary to spinal fusion anomalies. Neither two-dimensional ultrasound nor MRI identified the rib anomalies, nor did they allow for correct identification of the more compromised lung. Spondylocostal and spondylothoracic dysostoses involve rib deformities and distortion of the bony thorax that lead to pulmonary compromise. Correct prenatal diagnosis might not be made with standard fetal imaging. Three-dimensional ultrasound should be pursued (when available) upon recognition of a thoracic scoliosis to fully assess rib development.

Introduction

Spondylocostal and spondylothoracic dysostoses are characterized by vertebral anomalies and rib malformations of varying degrees (1-3). The thoracic distortion may compromise pulmonary development, and concomitant nonskeletal anomalies may be present (1). Fetal diagnosis of these disorders is important for family education and planning of delivery and postnatal care.

Case report

A 35-year-old G3P2 Mexican patient with a history of benign essential hypertension and gestational diabetes was referred to a tertiary care center and a children's hospital for the antenatal diagnosis of severe scoliosis in the second trimester. Her three pregnancies were with the same unre-

lated partner, and her two living children were healthy. The family history was negative for congenital anomalies.

At 16 weeks' gestational age, a serum screen was negative, with revised risks of less than 1 in 5,000 each for Down syndrome, trisomy 18, and spina bifida. The second trimester fetal assessment was initially performed by an outside medical center at 18 weeks, 5 days' gestational age, and was notable for significant scoliotic curvature of the thoracolumbar spine, without either spinal dysraphism or observed vertebral anomaly.

At this time, the patient was referred to our center for high-risk care. A repeat second-trimester anatomy scan performed at 21 weeks, 6 days' gestational age confirmed a 70- to 80-degree kyphoscoliosis involving the thoracolumbar spine (Fig. 1). No other anomalies were noted. Sonographically, the chest size and heart-to-lung ratio appeared normal. Fetal activity was normal, and the long bones were normal in size and morphology. The cord origin was velamentous. Following genetic counseling, amniocentesis was declined.

Further imaging by MRI for evaluation of the spinal cord and brain was pursued. The fetal MRI study was performed on a 1.5-Tesla Siemens magnet at 30 weeks' gestational age, using multiplanar, single-shot, fast-spin echo and fast imaging with steady state procession. The fetal MRI scan showed a normal brain and no cord abnormality. Severe lower thoracic scoliosis, approximately 80 degrees left convex, appeared secondary to segmentation fusion anomalies affecting at least two levels (Fig. 2). A mild, left, convex,

Citation: Chapman T, Done S, Goldberg M. Fetal diagnosis of spondylocostal dysplasia: Limits of conventional fetal ultrasound & MRI in diagnosing anomalies. *Radiology Case Reports*. [Online] 2011;6:450.

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Competing Interests: The authors have declared that no competing interests exist.

DOI: 10.2484/rcr.v6i1.450

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Figure 1. Fetus with spondylocostal dysplasia. Demonstrative sagittal view of the thoracic spine acquired during second-trimester ultrasound shows abnormal curvature of the spine. Posterior ribs do not appear fused. Segmentation-fusion anomalies were appreciable in the lower thoracic spine (not shown).

upper thoracic curve was also observed and felt to be secondary to a segmentation-fusion anomaly. Due to fetal motion, dysraphism could not be excluded. Finally, the left hemithorax was felt to be subjectively small, secondary to scoliosis. No rib anomaly was appreciable.

Delivery by cesarean section at 31 weeks was prompted by premature rupture of membranes and variable decelerations detected by fetal monitoring. A male infant was born with Apgar scores of 7 and 9 at 1 and 5 minutes. The infant was admitted to the neonatal intensive care unit for prematurity, kyphoscoliosis, respiratory support, and nutritional support. An echocardiogram showed a patent ductus arteriosus but was otherwise normal.

A skeletal fetal radiographic survey showed severe, long-segment, congenital levoscoliosis centered at the thoracolumbar junction and measuring approximately 70 degrees from T3-L2. Segmentation anomalies were noted throughout the entire thoracic spine. There were 13 ribs on the left and 11 ribs on the right, with medial bony fusion involving the right fifth through seventh ribs and the right eighth and ninth ribs. Multiple right pedicles were fused in the midthoracic spine at the levels of multiple left-sided hemivertebrae (Fig. 3). The right lung was somewhat smaller and less aerated than the left. Five lumbar and five sacral vertebrae were normally formed. The skull and long bones were normal.

Discussion

This case of spondylocostal dysplasia illustrates the limitations of conventional prenatal imaging in a full evaluation of the bony thorax. Although the mother's prenatal care was notable for advanced maternal age, essential hypertension, and gestational diabetes, there was no family history of vertebral anomalies or scoliosis. The affected fetus was diagnosed in the second trimester with a thoracolumbar

kyphoscoliosis by ultrasound. A third-trimester fetal MRI study successfully identified vertebral anomalies as the cause of a severe thoracolumbar scoliosis but, like the fetal ultrasound studies, failed to reveal the rib anomalies.

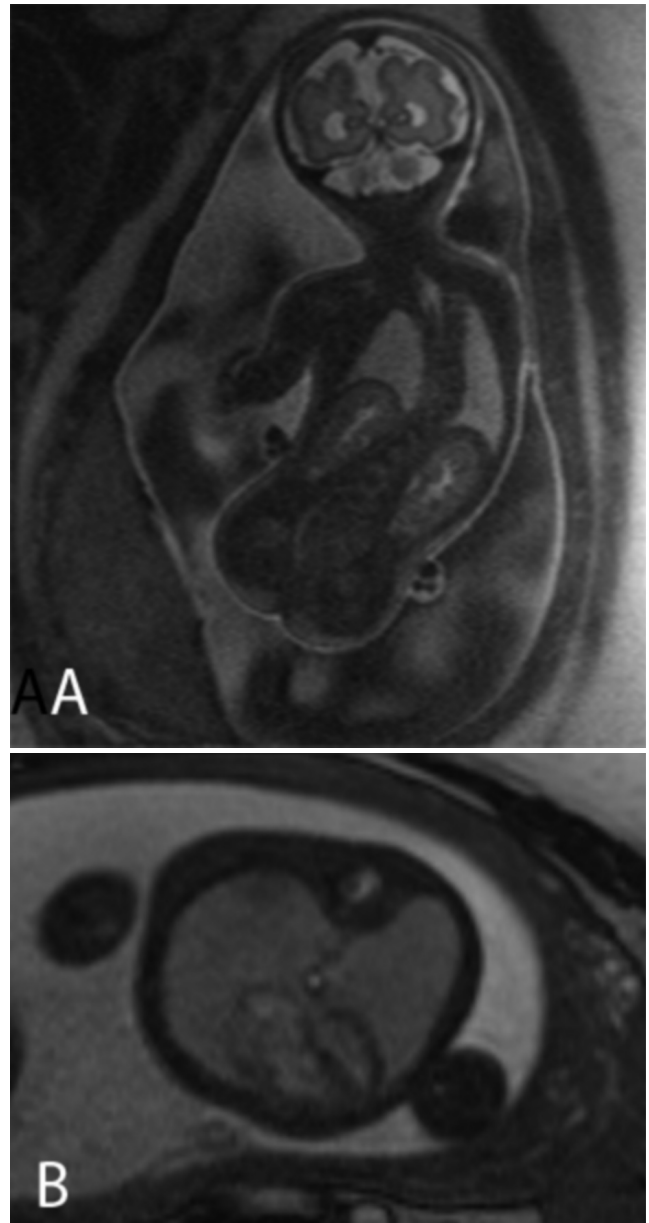


Figure 2. Fetus with spondylocostal dysplasia. Coronal HASTE (A) and axial FISP (B) images from a third-trimester fetal MRI study show a lower thoracic left-convex curve. The left lung volume appeared subjectively smaller on the fetal MRI study. Neither T2-weighted sequence elucidated rib anatomy.

Isolated congenital scoliosis is not as complex a diagnosis as spondylothoracic dysplasia, which includes rib anomalies such as posterior fusion, aplasia, and irregular rib shape

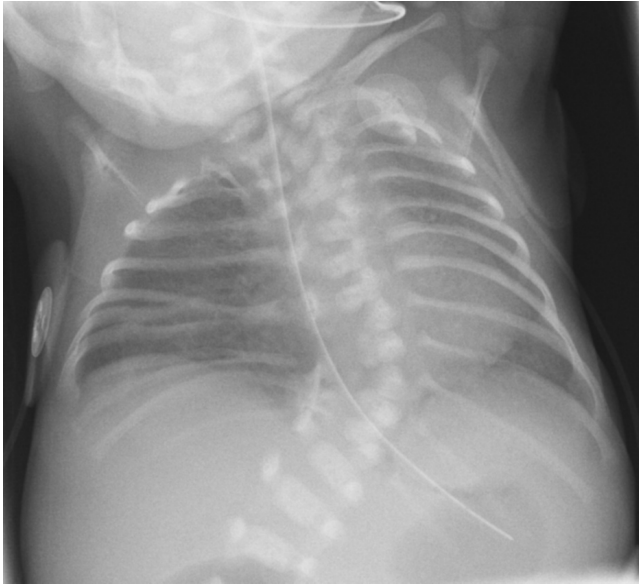


Figure 3. Fetus with spondylocostal dysplasia. Postnatal chest radiograph shows severe left-convex scoliosis centered at the thoracolumbar junction and measuring approximately 70 degrees from T3-L2. Segmentation anomalies appear throughout the entire thoracic spine. There are 13 ribs on the left and 11 ribs on the right, with medial bony fusion involving the right fifth through seventh ribs and the right eighth and ninth ribs. The right lung is somewhat smaller and less aerated than the left.

and size. The small chest and inadequate lung growth leads to thoracic insufficiency syndrome. While respiratory support and chest physiotherapy are the hallmarks of care at birth (1), for many children with progressive scoliosis and respiratory insufficiency syndrome, surgical intervention is often needed later. Growing-rod techniques are recommended for progressive scoliosis; in children whose primary problem is rib fusions and scoliosis; however, expansion thoracostomy and vertical expandable prosthetic titanium rib (VEPTR) are preferred (4).

Routine second-trimester fetal imaging includes a systematic evaluation of the skeletal axis, thorax, and extremities (5, 6). Detection of small osseous abnormalities such as hemivertebrae requires meticulous examination of the fetus using coronal and sagittal planes during ultrasound (7, 8). Three-dimensional and four-dimensional ultrasound offer additional value for evaluation of the spine (9, 10) but are not widely available. Because of these limitations and because of the inherent limitations of MRI in the evaluation of bone, low-dose computed tomography is being explored at some centers for further clarification of fetal skeletal dysplasia cases (11).

In cases of bony thoracic distortion or short ribs, the lung volumes must be assessed, most readily by measuring the chest circumference at the level of the four-chamber view of the heart and comparing this to gestational age standards. Pulmonary hypoplasia is suspected if circumference is

less than the 5th percentile (6). Many other methods have been standardized and include comparison of chest circumference to that of the abdomen and to the trunk length. Similarly, assessment of lung volumes by fetal MRI may be done qualitatively or quantitatively, and is nicely reviewed by Deshmukh et al (12).

In the case presented here, fetal ultrasound showed the ratio of the heart area relative to the chest area to be normal. The affected fetal left lung appeared more compromised on the left side by MR imaging. However, postnatal chest radiographs showed a smaller right lung than left lung. Because of the premature delivery, the neonatal ICU team was prepared at the time of cesarean delivery. Under other circumstances, without prenatal anticipation of pulmonary compromise, immediate postnatal intensive care might have been delayed. Therefore, when fetal imaging suggests a complicated scoliosis, evaluation of the ribs is important. Two-dimensional ultrasound and MR imaging may be inadequate. More in-depth imaging by three-dimensional ultrasound should be considered.

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Fetal diagnosis of spondylocostal dysplasia: Limits of conventional imaging in diagnosing anomalies

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