Sonographic Diagnosis of Caudal Regression in the First Trimester of Pregnancy

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Caudal regression syndrome (CRS) represents a continuum of congenital anomalies that may include incomplete development of the sacrum and, to a lesser extent, the lumbar vertebrae, disruption of the distal spinal cord, and extreme lack of growth of the caudal region.[1] First-trimester prenatal diagnosis of CRS is challenging because of incomplete ossification of the sacrum at that time.[2] Most case reports of CRS have occurred during the second trimester.[3–10] We report a case of CRS diagnosed during the first trimester on the basis of transvaginal sonography.

Abbreviations
CRS, caudal regression syndrome

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Case Report

A 26-year-old gravida 2, para 1 woman was seen in our emergency department with a 2-day history of nausea and vomiting. Her last menstrual period was uncertain, but by pelvic examination she was found to be about 10 weeks' pregnant. The patient had a 4-year history of non–insulin-dependent diabetes mellitus without adequate control. Maternal hyperglycemia and ketonuria prompted admission for suspected diabetic ketoacidosis.

During the admission, sonography was performed for appropriate dating of pregnancy. This study was performed transvaginally, which revealed a singleton intrauterine pregnancy at 10.9 weeks by crown-rump length measurement (40 mm) and fetal heart activity. Absence of the lower spine and lower limb abnormalities were suspected, indicative of CRS (Figs. 1 and 2). The glycosylated hemoglobin value was 9.8%. The parents were counseled at length about the sonographic findings and the prognosis of the pregnancy. They opted for medical

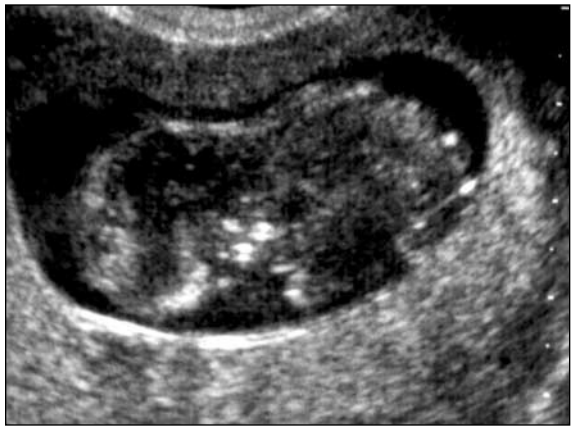
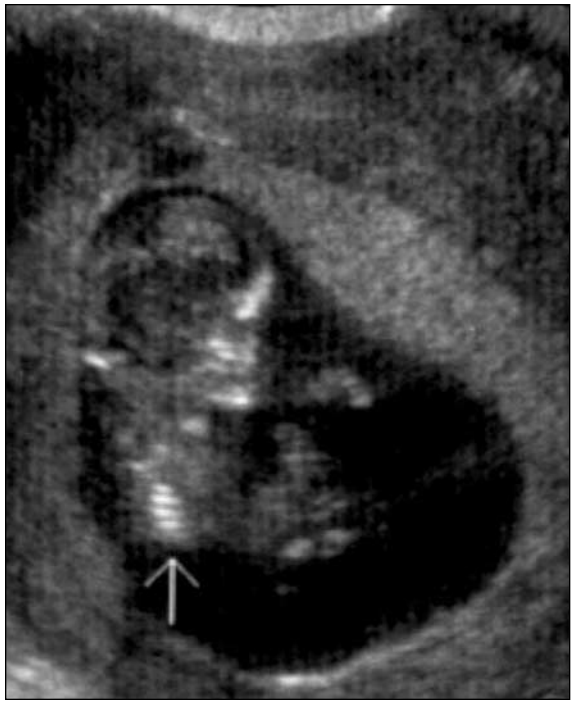


Figure 1. Transvaginal sonogram showing absence of the lower spine.

termination of the pregnancy with misoprostol. Pathologic evaluation revealed a female fetus with a weight of 14 g and a crown-heel length of 7.8 cm. consistent with 12.5 weeks' gestation. The vertebral column was missing from T11 to the sacrum (Fig. 3). All other organs examined, including the kidneys, were normal. Radiologic studies of the fetus confirmed the sonographic findings (Fig. 4).

Figure 2. Transvaginal sonogram showing lower limb abnormalities.

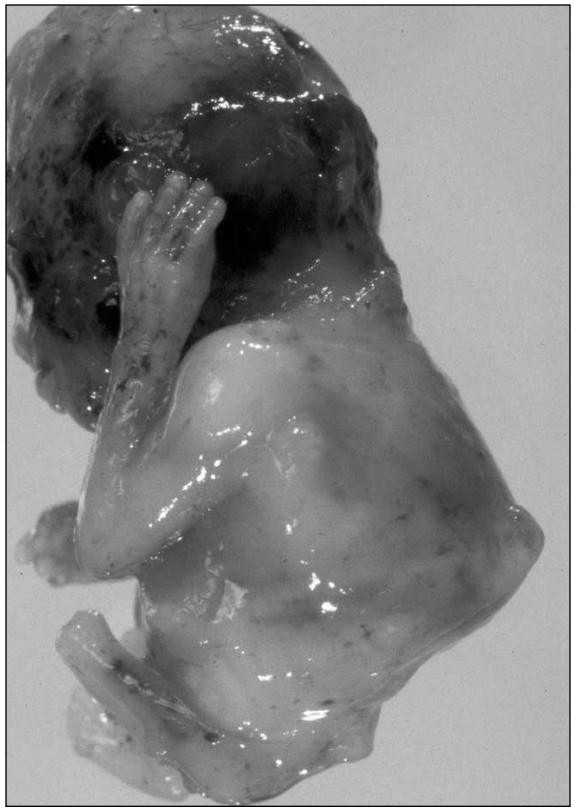


Discussion

Caudal regression syndrome is a rare anomaly, with 300 cases thus far repoted.[10] Differentiation of the lower spine is usually complete before the seventh week of pregnancy. Previously grouped with sirenomelia as the most severe form, it is now thought that the two are pathogenetically unrelated.[1] Approximately 16% of cases are seen in infants of diabetic mothers.[2] The etiology is unknown, and although usually sporadic, a few instances of affected parents have been described.[1]

This report represents an early prenatally diagnosed case of CRS. Detailed evaluation of the fetal spine and lower extremities are an essential part of every obstetric sonographic examination. Multiple views in the sagittal, transverse, and coronal planes allow for the early detection of spinal anomalies associated with this syndrome. The transvaginal approach has added even more to the possibilities that sonologists can diagnose rare anomalies such as CRS at an earlier gesta-

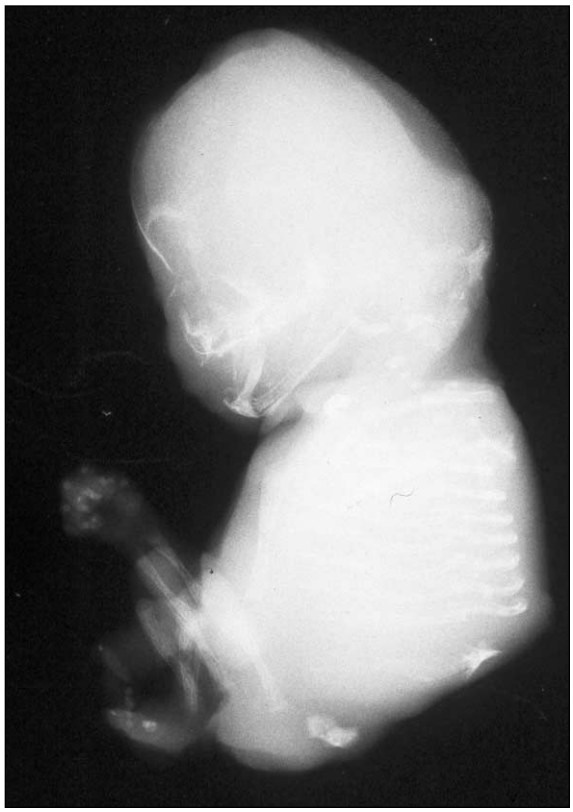
Figure 3. Lateral photograph of the fetus. Note the malformation beyond the thoracic spine.



tional age than previously reported. A MEDLINE search showed that all previously reported cases had a gestational age range of 11 to 23 weeks. Baxi et al[4] reported a case in which at 9 weeks' gestation a shortening of the crown-rump length and a protuberance of the lower spine suggested CRS, but the diagnosis was not made with certainty until 17 weeks. Recently, Fukada et al[10] reported a case of CRS prenatally diagnosed after detection of a large nuchal translucency, but the diagnosis of CRS was not made until 16 weeks.

Sonographic findings are variable and will depend on the extent and severity of the defect. Diagnosis is usually made by visualizing a shortened spine with missing sacral and lower lumbar vertebrae. Vertebrae are not shown at the L5 level. Associated anomalies are common, most frequently involving the genitourinary, gastrointestinal, and cardiovascular systems. Detailed obstetric sonography and echocardiography are recommended as part of the workup after a diagnosis of CRS is made.

Figure 4. Lateral radiograph of the fetus. Note the absence of the lumbar spine and sacrum.



Because this syndrome is not associated with aneuploidy, a fetal karyotype is not warranted. The prognosis will depend on the severity of the defect and presence of associated abnormalities.

Early diagnosis of this rare entity, which has a high morbidity rate, gives parents time to make informed decisions that at a later gestational age may not be available. This case raises the important issue of early screening for anomalies in high-risk patients, such as those with pregestational diabetes and poor glycemic control during organogenesis.

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