High-Resolution Sonographic Diagnosis of Sirenomelia

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Sirenomelia sequence is a rare lethal pattern of congenital anomalies characterized by a number of hallmark skeletal anomalies, including fusion of the lower extremities or a single lower limb, bilateral renal agenesis or dysgenesis with absent or hypoplastic renal arteries, oligohydramnios, and the presence of aberrant vasculature. The characteristic feature of sirenomelia consists of the varying degree of fusion of the lower limbs. Two separate systems of classification of sirenomelia have been proposed, based on the number of feet or the pattern of fused bones. Prenatal diagnosis of this condition can be accomplished by sonography and relies on the inability to show separate lower limbs or the identification of a single femur. However, a confident diagnosis is usually difficult during the second trimester because of the severe oligohydramnios associated with the condition, which hampers a proper evaluation of fetal lower extremities. This can be overcome by amnioinfusion, but it is an invasive method. Alternatively, magnetic resonance imaging can be used because it is less hampered by oligohydramnios, but it is not easily available. Recently, color Doppler sonography has been reported to be useful in the diagnosis. It reveals the origin of the aberrant vitelline artery that continues as the single umbilical artery. The distal aorta may be atretic or hypoplastic. The iliac and femoral arteries may not be seen. However, this method also faces difficulty if the femoral arteries are visualized. Here we report the use of high-resolution sonography as an easy and simple technique for the diagnosis of the sirenomelia in the presence of oligohydramnios.

The patient was a 22-year-old primigravida in her 30th week of gestation. Sonography revealed a gestation of 29 weeks with anhydramnios. The fetus showed features of bilateral renal agenesis and a single umbilical artery, which suggested the possibility of sirenomelia sequence.
Two separate femurs were seen (Figure 1). Because of the anhydramnios, it was not possible to rule out soft tissue fusion of the lower limbs. The color Doppler study was not diagnostic because the aorta was seen bifurcating into the iliac arteries (Figure 2A). Both femoral arteries were seen in the thigh (Figure 2B). A linear 5- to 12-MHz probe was used, which revealed the continuous skin line over both femurs on a transverse scan of the thigh (Figure 3), indicating fused thighs. The soft tissue fusion of the thighs was also visible on a coronal scan of the thighs. The echogenic skin lines between the 2 thighs that would be expected in separate thighs were absent (Figure 4). The bones of the legs were not seen except for a short tibia, distal to the fused thighs. These features were diagnostic of sirenomelia. The pregnancy was terminated. The abortus showed the fused thighs of sirenomelia (Figure 5).
In conclusion, high-resolution sonography is an easy alternative technique that can be used even in the presence of anhydramnios for the diagnosis of sirenomelia. It allows direct visualization of the abnormalities of sirenomelia by depicting the continuous skin over the 2 femurs in the fused lower limbs.

References


