

Case report: Sirenomelia-early second trimester antenatal ultrasonographic diagnosis

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Sirenomelia is a rare lethal fetal anomaly that can be accurately diagnosed in the late first trimester or in the early second trimester with USG. Reports of antenatal diagnosis before 20 weeks of gestation are very rare in the literature.^[1] One case of sirenomelia diagnosed at 18 weeks of gestation is described and discussed briefly. An early diagnosis with antenatal USG helps to plan treatment.

Case Report

A 22-year-old primigravida with 18 weeks of amenorrhea was referred for routine antenatal USG. Her personal and family history were unremarkable. There was no history of any medication or infection in the early period of pregnancy. The scanning was performed using a 3.5–5 MHz high-definition transabdominal transducer and a 6.5–10 MHz endovaginal transducer (Aloka Prosound 5000). This showed an active fetus surrounded by moderate volume of liquor. The placenta, head, upper trunk, upper vertebrae, and the stomach shadow were normal. The renal and urinary bladder shadows were not discerned.

The lumbar and sacrococcygeal vertebrae were very poorly formed and were disfigured. Distal to this, the pelvic bones were represented by specks of calcification [Figure 1]. The femora were fused proximally and forked distally [Figure 2]. Two long bones were seen distally, representing the tibiae, with surrounding muscle mass. The rudimentary feet appeared as a small irregular soft tissue mass with a dense echoic speck at the distal end of the fused limbs [Figure 3].

These USG features were consistent with a diagnosis of sirenomelia. The parents opted for therapeutic abortion. The abortus showed the typical morphological features of sirenomelia.

Discussion

Sirenomelia (also known as mermaid deformity, symmelia,

sympus, symposia, uromelia, and monopodia) is an invariably lethal anomaly with a reported incidence of 1 in 60, 000–100,000 live births.^[1] Around 300 have been reported in the world literature, of which 13 cases have been from India.^[2–4] Most of them were diagnosed after

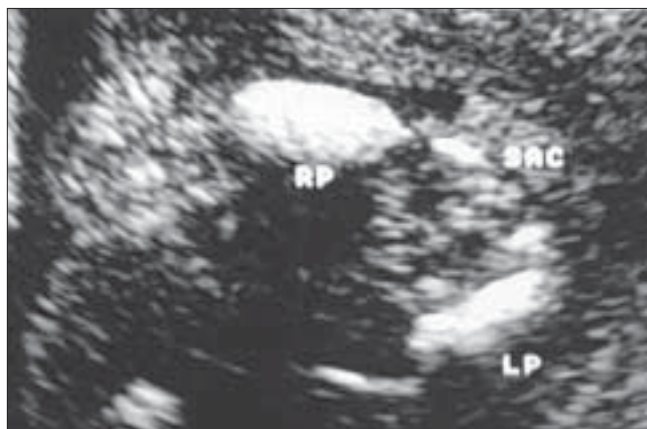


Figure 1: An axial scan at the level of the pelvis shows sacrococcygeal dysgenesis (SAC) and deformed pelvic bones (RP, LP)

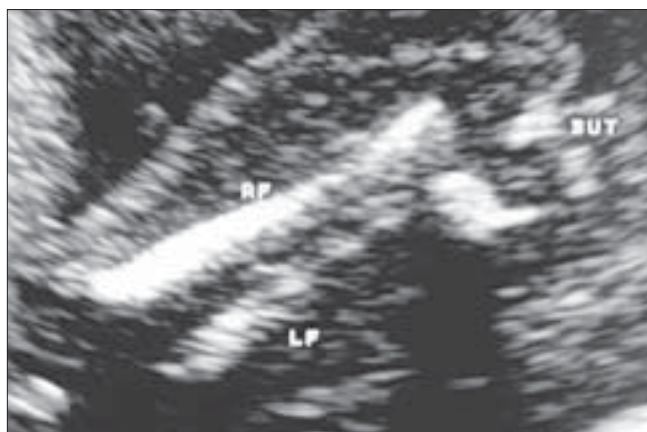


Figure 2: The two femora (RF, LF) are fused proximally and forked distally. The soft tissue shadow around these bones forms the proximal common thigh

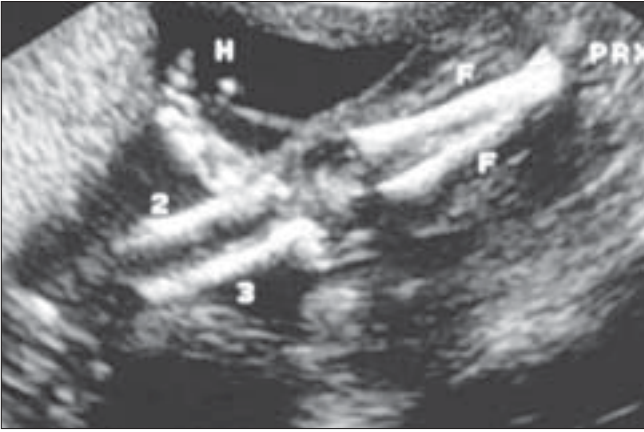


Figure 3: The two tibiae (2, 3) are lying parallel to each other forming a common leg. The distal echogenic area represents the rudimentary fused feet. (F-proximally fused femora, PRX- proximal end of fused femora, H-part of normal hand)

birth and no definite antenatal diagnosis was possible in all of these cases.^[2-4]

This anomaly is characterized by varying degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary atresia, and anorectal atresia.^[1] The generally accepted theory—the vitelline artery steal theory—attributes the anomaly to the presence of an aberrant arterial shunt from the upper abdominal aorta, which diverts blood from the lower torso to the placenta, leading to a hypoplastic lower aorta and maldevelopment of the lower torso.^[5]

The fetus illustrated here had all the typical features of sirenomelia. The oligohydramnios was less severe than what is usually reported, as the fetal age was only 18 weeks at the time of scanning. The oligohydramnios worsens when the contribution from fetal urine is jeopardized in late pregnancy. A definitive diagnosis is, therefore, usually difficult because of poor visualization of fetal details due to oligohydramnios in the late trimester. At these times, a high-resolution probe may define the continuity of the skin contour over the fused limbs,^[6] thus helping to diagnose sirenomelia. Color Doppler imaging can add to the diagnostic accuracy if the abnormal fetal vasculature is represented by a single umbilical artery in association

with bilateral renal agenesis.^[7] In a recent report, the authors diagnosed sirenomelia at 10 weeks of gestation and confirmed it at 15 weeks.^[8] The fetus in our report, at 18 weeks, showed proximally fused femora and separate tibiae. The fibulae were absent. The differential diagnoses include caudal regression sequence, especially among diabetic mothers, sacral agenesis like cloacal extrophy and the VACTERL association (vertebral defects, imperforate anus, cardiac anomalies, tracheo–esophageal fistula, radial and renal dysplasia, and limb anomalies).^[8,9] Recent advances in the understanding of embryonic axial mesoderm patterning suggest that sirenomelia represents the most severe manifestation of the caudal regression spectrum, which also shows similar features.^[9]

The importance of an early antenatal USG is highlighted by the fact that this is an invariably lethal condition and therapeutic abortion can be carried out earlier.

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