# First Trimester Diagnosis of Sirenomelia by Real-Time Three-Dimensional Ultrasound: Case Report

## Gerçek-Zaman Üç-Boyutlu Ultrasonografi ile İlk Trimester Sirenomeli Tanısı

Gökalp ÖNER, MD,[a]
Cem BATUKAN, MD,[a]
Mahmut Tuncay ÖZGÜN, MD,[a]
Mustafa BAŞBUĞ, MD[a]

[a]Department of Obstetrics and Gynecology,
Erciyes University Faculty of Medicine, Kayseri,

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Yazışma Adresi/*Correspondence:*
Gökalp ÖNER, MD
Erciyes University Faculty of Medicine, Department of obstetrics and Gynecology, Kayseri, TÜRKİYE/TURKEY
dr.gokalpw85@mynet.com

**ABSTRACT** Sirenomelia, characterized by complete fusion of the lower limbs in combination with severe urogenital and gastrointestinal malformations and it is a rare and usually lethal disorder. The incidence is about 0.01 to 0.16 per 10000 live births and it is three times more common in male fetuses than females. The prenatal diagnosis of malformations with three-dimensional sonography could increase the physician's confidence and understanding of the fetal condition which was found by two-dimensional ultrasound. We report a case of sirenomelia detected by two- and three dimensional ultrasound at 11 gestational weeks. Although the prenatal diagnosis is usually made early in the second trimester of pregnancy with oligohydramnios as the alerting sign, sirenomelia should be carefully identify in the first trimester when an intraabdominal cystic structure is detected on prenatal ultrasound.

**Key Words:** Ectromelia;prenatal diagnosis; ultrasonography prenatal

**ÖZET** Sirenomeli, ağır ürogenital ve gastrointestinal malformasyonlarla kombine olmuş alt ekstremitelerin tamamen birleşmesiyle karakterize, nadir görülen ve sıklıkla öldürücü bir hastalıktır. İnsidansı yaklaşık olarak 10.000 canlı doğumda 0.01 ile 0.16 arasındadır, erkek fetuslar dişilere oranla üç kat daha fazla etkilenmiştir. Malformasyonların üç boyutlu ultrasonografi ile prenatal tanısı, iki boyutlu ultrasonografi ile bulunmuş fetusa ait durumun anlaşılmasını ve hekimin itimatını arttırabilmektedir. Biz 11. gestasyonel haftada iki ve üç boyutlu ultrasonografi ile tespit edilmiş bir sirenomeli olgusu sunduk. Prenatal tanısı, uyarıcı işaret olarak oligohidroamniyozla sıklıkla erken olarak ikinci trimesterde konulmasına karşın, sirenomeli prenatal ultrasonografide intraabdominal kistik yapı saptandığında ilk trimesterde dikkatlice tanınabilmektedir.

**Anahtar Kelimeler:** Ektromeli, prenatal tanı, ultrasonografi prenatal

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Sirenomelia, characterized by complete fusion of the lower limbs in combination with severe urogenital and gastrointestinal malformations, is a rare and usually lethal disorder.[1] Progressive oligohydramnios due to agenesis or dysgenesis of the kidneys is usually the first sign of this lethal malformation during the second trimester. Early prenatal diagnosis of sirenomelia can be achieved by sonographic demonstration of constant simultaneous movements of the lower limbs and/or the identification of a skeletal anomaly like a single femur.[2]

This report provides a case of sirenomelia detected by using three-dimensional (3D) ultrasound at 11th gestational weeks.

## CASE REPORT

A 22-year-old woman, gravida 3, para 1, was referred for a targeted ultrasound investigation to our ultrasound unit in the 11th week of gestation because an intraabdominal cyst was detected during a routine scan. B-mode transabdominal and transvaginal ultrasound examinations showed a left-sided intraabdominal cystic structure measuring 9.6 x 9.4 mm in diameter, which was located just beneath the stomach (Figure 1). Meticulous scanning revealed fusion of the lower limbs, an abnormal pelvic bone and a single umbilical artery. The sacrum, both kidneys and bladder were also not visible. These sonographic findings favored the diagnosis of sirenomelia. Presence of lower extremity fusion was further supported by transabdominal 3D ultrasound imaging (Figure 2). After non-directive counseling, pregnancy termination was requested by the couple. Postmortem radiological study revealed a single femur, absent sacrum, and hypoplastic pelvic bones, which is consistent with the prenatal diagnosis (Figure 3). The cystic structure detected on prenatal ultrasound turned out to be dilatation of sigmoid colon during autopsy and the gross appearance of the fetus confirmed the existence of a single lower extremity (Figure 4). A scrupulous dissection showed bladder agenesis and presence of intraabdominally located gonads. Placental examination revealed a single umbilical artery. Fetal karyotype was 46, XX.

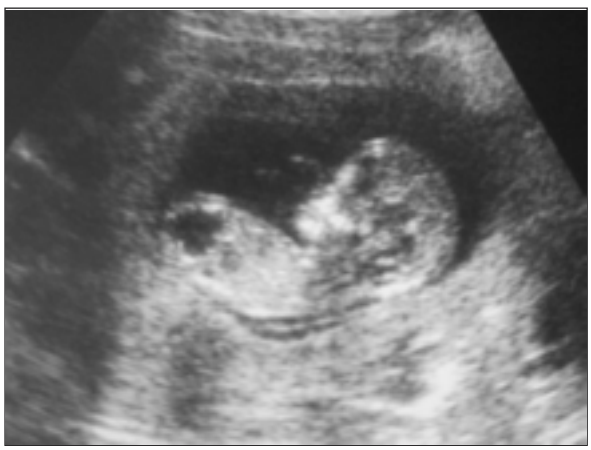

**FIGURE 1:** Two-dimensional ultrasound scan at 11 weeks of gestation: intraabdominal cystic structure is in the forefront, demonstrating the urogenital and/or gastrointestinal malformation.

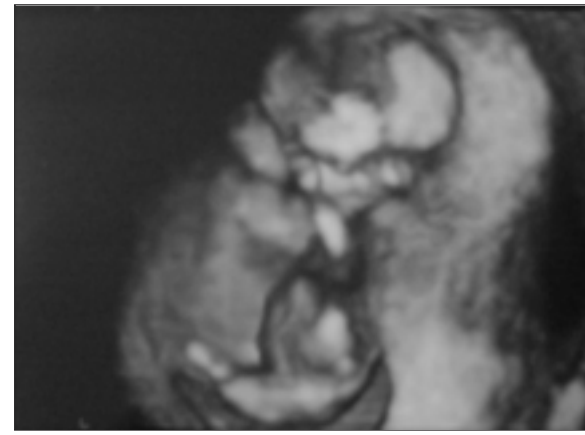

**FIGURE 2:** 3D ultrasound image of the sirenomelia with fusion of the lower extremities.

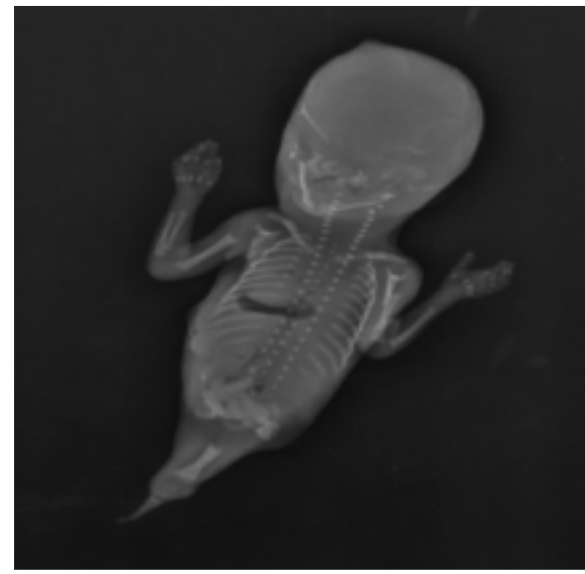

**FIGURE 3:** Postmortem X-ray.

## DISCUSSION

Sirenomelia sequence combines in all cases fusion and rotation of the lower limbs to various degree and anorectal atresia. Renal agenesis or dysplasia, bladder agenesis and absence of internal and external genitelia except the gonads are noted in nearly all cases. Dysgenesis or even agenesis of the sacrum may be associated with extensive vertebral dysgenesis. The early fusion of the lower extremities confers a siren-like appearance of the fetus.[3] The incidence is about 0.01 to 0.16 per 10000 live births, males are three times more often affected than females and the incidence is higher in one of the iden-

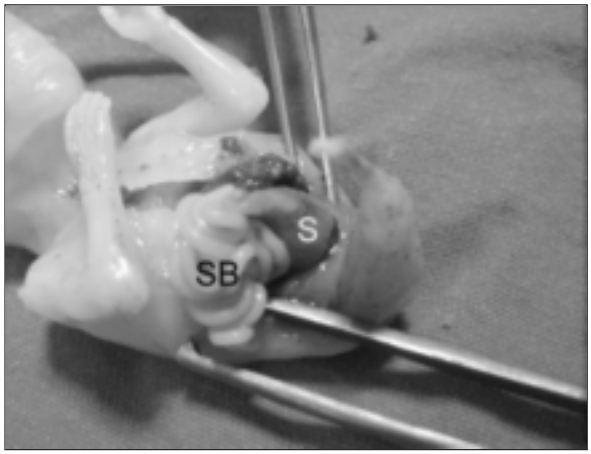


**FIGURE 4:** Autopsy showing normal small bowel (SB) and dilatation of the sigmoid colon (S).

tical twins.[4] The etiology of sirenomelia is not clear, however, vascular disruption or caudal embryo damage was proposed. Additionally, chromosomal abnormalities are rare in cases with sirenomelia.[5]

Although urinary tract dysgenesis or agenesis appears to begin from the end of the first trimester, most cases of sirenomelia were diagnosed early in the second trimester due to oligohydramnios as an alerting sign to the sonographer.[6,7] As during the early weeks of gestation the amniotic fluid is mainly secreted by the placenta and the umbilical cord, amniotic volume may not be helpful for the prenatal diagnosis of sirenomelia in the first trimester.

In our case, the cystic structure, which was detected at 11 weeks of gestation, reflects the complex nature of sirenomelia, which comprises malformations of the lower abdomen, urogenital system and intestine. Dilatation of the sigmoid colon due to an imperforate anus has been reported as an intraabdominal cystic structure in previous cases with first trimester diagnosis of sirenomelia.[5] Therefore this sonographic finding might be an important indirect sign of serious lower extremity abnormalities, especially sirenomelia.

The differential diagnosis of abdominal cysts during the first trimester includes megacystis, although this is unlikely at 11 weeks of gestation.

3D ultrasound images of two cases with sirenomelia diagnosed in the first trimester were firstly provided by Monteagudo et al, which aided to further characterize the conventional 2D sonographic findings.[8] The prenatal diagnosis with 3D sonography were thought to be the additional information obtained namely was the overall view, the confirmation of 2D findings, and the abnormal fetal movements, which could increase the physician's confidence and understanding of the fetal condition.[9] Although 3D ultrasound offers diagnostic advantages for visualising skeletal anomalies, 2D ultrasound presents a better view for the intraabdominal cystic structure. An early antenatal diagnosis of sirenomelia may be suspected in the presence of bilateral renal agenesis, malformed lower limbs and a single umbilical artery.[5] Such sonographic signs were readily apparent with 2D ultrasound examination.

In conclusion, sirenomelia should be suspected when an intraabdominal cystic structure is visible during a routine first trimester ultrasound scan. Subsequent 3D ultrasound may be helpful in confirming the diagnosis of a sirenomelia.

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