# Prenatal Diagnosis of Sirenomelia

**M. Aykut ÖZEK[1], Mert TURĞAL[1], Özgür ÖZYÜNCÜ[1], M. Sinan BEKSAÇ[1]**

*Ankara, Turkey*

Sirenomelia is a rare and lethal congenital malformation. It is associated with a variety of anomalies and oligo/anhydramniosis. We present a case of sirenomelia detected in early second trimester presenting with anhydramniosis.

**Key Words:** Sirenomelia, Anhydramniosis, Prenatal diagnosis

Gynecol Obstet Reprod Med 2013;19:36-38

## Introduction

Sirenomelia is also known as "mermaid syndrome," because of the one lower extremity. It is a very rare malformation that is incompatible with life and an estimated incidence of the sirenomelia is 1:60000.[1] Sirenomelia is the most severe form of caudal regression syndrome, in which the two lower limbs are fused in a single abnormal limb. Sirenomelia sequence is associated with anorectal malformation, fusion of inferior limbs, renal agenesis, severe vertebral anomalies, genital anomalies.[2,3]

Prenatal diagnosis of the sirenomelia may be easier make during the first trimester than the second trimester, because amniotic fluid is relatively normal. In this period, source of amniotic fluid is amniotic membrane covering placenta and umbilical cord. Ultrasonographic diagnosis of the sirenomelia in second trimester may be prevented by severe oligohydramnios the, which is caused by renal agenesis or dysgenesis. Most cases are diagnosed postnally.

## Case Report

A 27-year-old woman, gravida 2 para 0 was referred to Perinatology Unit of our hospital at 17 weeks of gestation for evaluation of severe oligohydramniosis. Her previous pregnancy was terminated because of anhydramniosis at 17$^{th}$ gestational week. She had an operation for atrial septal defect; and she is otherwise healthy. She and her husband were not relatives. She did not report any history of exposure to a teratogen.

Sonography revealed a single live fetus with anhydramnios. Fetal biometry was normal. Kidneys were dysplastic bilaterally and lateral ventricles were dilated minimally (atrial diameter was 8.2 mm). Due to absent amniotic fluid, anatomic survey was very difficult to perform. The parents were informed of the poor prognosis of anhydramnios at that age of gestation and they elected termination of pregnancy. Pregnancy was terminated with misoprostol induction and a fetus weighting 120 g was delivered. We postnatally observed that the lower limbs and feet of the fetus were fused (Figures 1A and 1B). The parents opted for a full autopsy. Autopsy revealed findings consistent with sirenomelia.

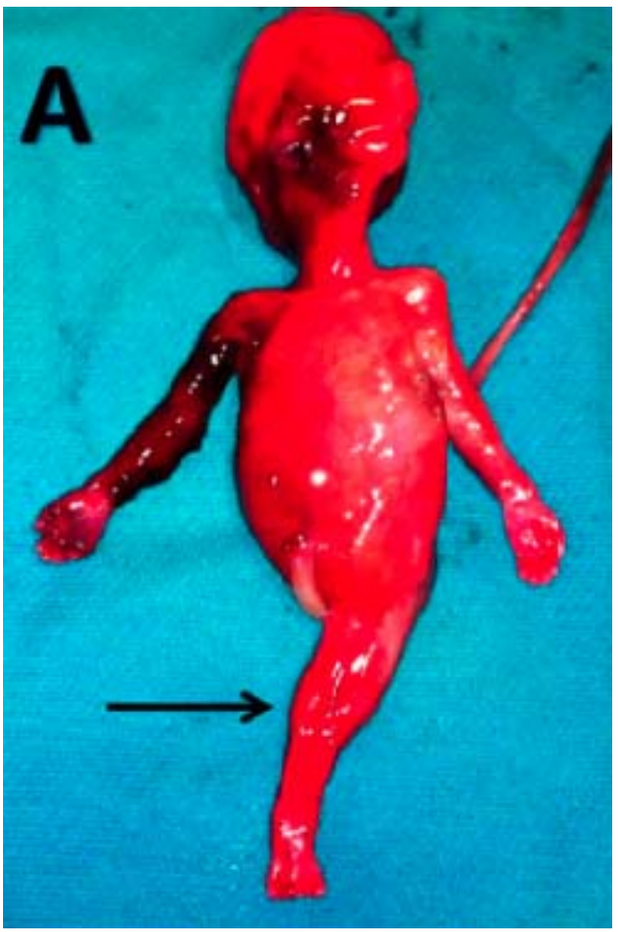


*Figure 1A: Macroscopic features of the affected fetus A: Black arrow indicating fused lower extremities*

*[1]Hacettepe University Medical Faculty Department of Obstetrics and Gynecology, Ankara*

*Address of Correspondence:* *M. Aykut Özek*
*Hacettepe University Medical Faculty,*
*Department of Obstetrics and*
*Gynecology, 06100, Sıhhiye, Ankara*
*aykut.spm@gmail.com*

*Submitted for Publication:* *03. 03. 2013*
*Accepted for Publication:* *15. 04. 2013*

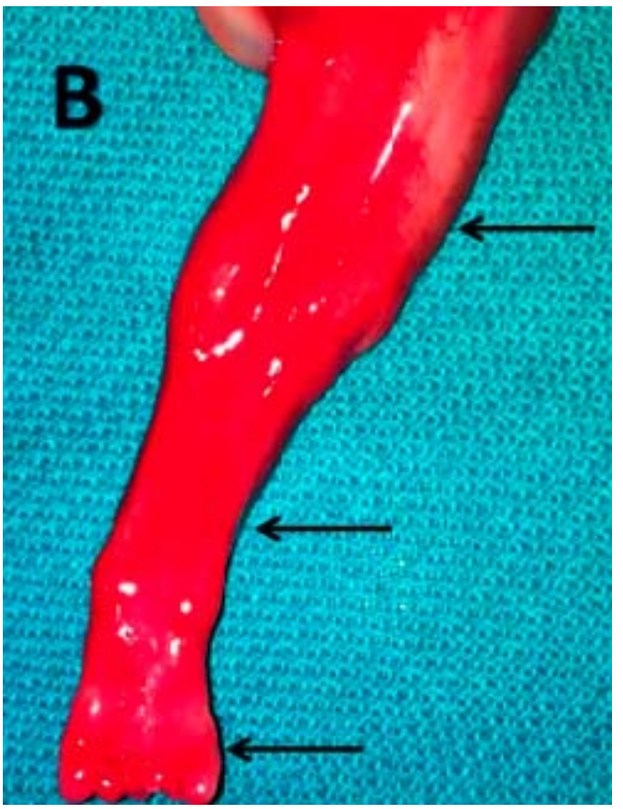


*Figure 1B: Magnified view of the fetus, black arrows indicating fused long bones and feet*

## Discussion

Sirenomelia takes its name from the siren-like appearance of the fusion of the lower extremities. The sequence is very rare with an estimated incidence of 1 per 60,000 births.[1] It affects predominantly male fetuses with a sex ratio of 2,7:1.[4] It occurs 100 to 150 times more frequently in monozygotic twins than dizygotic twins or singletons.[5] This malformation is mostly lethal, survival is very rare in literature.[6]

The etiology is controversial but there are hypotheses about vascular disruption and caudal embryo damage.[2] Other theories of teratogenic exposure like retinoic acid, cocain and association with diabetes mellitus have been described based on cases.[7-10]

Sirenomelia sequence may be divided into three groups based on the number of the feet present: symelia apus (the most common type, feet are absent), symelia unipus (presentation of one foot) and symelia dipus (two distinct feet but are malrotated).[7] According to Stocker's classification, seven variants may be identified.[3]

The sirenomelia sequence combines in all cases anorectal atresia and fusion and rotation of the lower limbs. Urinary tract anomalies consisting of renal agenesis or dysplasia and bladder agenesis; absence of the internal and external genitalia except the gonads are noted nearly in all cases.[2,3]

Antenatal diagnosis of sirenomelia is difficult. It is usually suspected during second trimester with oligohydramnios related to renal dysgenesis or agenesis. But in this setting, severe oligohydramnios makes the morphological assessment of the fetus to become more difficult. Therefore the diagnosis is usually made at autopsy. Amnioinfusion may be used to improve sonographic assessment.[11] MRI also can be utilized because it is not affected from the diminished amniotic fluid.[12] During first trimester, the primary source of amniotic fluid is amniotic membrane, so first trimester or early second trimester diagnosis of this rare sequence is preferable.[4]

Early diagnosis of sirenomelia is suspected in the presence of fused lower extremities, bilateral renal agenesis or dysgenesis and single umbilical artery deriving from a persistent vitelline artery. Color and power Doppler ultrasound was performed successfully and revealed the aberrant vasculature in these fetuses.[13] The main differential diagnosis is bilateral renal agenesis and in these cases, Doppler ultrasound can be confidently used to differentiate this condition with other causes of renal agenesis.[14]

In conclusion, since it is almost invariably lethal, early diagnosis and termination of pregnancy at early weeks' of gestation is important.


## Sirenomelide Prenatal Tanı

Sirenomeli nadir ve ölümcül bir konjenital malformasyondur. Çeşitli anomaliler ve oligo/anhidramniyozis ile ilişkilidir. Bu olgu sunumunda, merkezimize erken ikinci trimesterde anhidramniyozis ile başvuran bir sirenomeli vakasını sunuyoruz.


**Anahtar Kelimeler:** Sirenomeli, Anhidramniyozis, Prenatal tanı

## References


1 Nyberg DA, Mahony BS, Pretorius D: Diagnostic ultrasound of fetal anomalies: Text and atlas. Chicago, Year Book Medical Publishers 1990.

2 Stevenson RE, Jones KL, Phelan MC, Jones MC, Barr M, Jr., Clericuzio C, Harley RA, Benirschke K: Vascular steal: The pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics 1986;78:451-7.

3 Stocker JT, Heifetz SA: Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol 1987;10:7-50.

4 Akbayir O, Gungorduk K, Sudolmus S, Gulkilik A, Ark C: First trimester diagnosis of sirenomelia: A case report and

review of the literature. Arch Gynecol Obstet 2008; 278:589-592.

5 Kapur RP, Mahony BS, Nyberg DA, Resta RG, Shepard TH: Sirenomelia associated with a "vanishing twin". Teratology 1991;43:103-108.

6 Schiesser M, Holzgreve W, Lapaire O, Willi N, Luthi H, Lopez R, Tercanli S: Sirenomelia, the mermaid syndrome--detection in the first trimester. Prenat Diagn 2003;23: 493-5.

7. Sarpong S, Headings V: Sirenomelia accompanying exposure of the embryo to cocaine. South Med J 1992; 85:545-7.

8 Von Lennep E, El Khazen N, De Pierreux G, Amy JJ, Rodesch F, Van Regemorter N: A case of partial sirenomelia and possible vitamin a teratogenesis. Prenat Diagn 1985;5:35-40.

9 Twickler D, Budorick N, Pretorius D, Grafe M, Currarino G: Caudal regression versus sirenomelia: Sonographic clues. J Ultrasound Med 1993;12:323-30.

10 van Zalen-Sprock MM, van Vugt JM, van der Harten JJ, van Geijn HP: Early second-trimester diagnosis of sirenomelia. Prenat Diagn 1995;15:171-77.

11 Blaicher W, Lee A, Deutinger J, Bernaschek G: Sirenomelia: Early prenatal diagnosis with combined two- and three-dimensional sonography. Ultrasound Obstet Gynecol 2001;17:542-3.

12 Van Keirsbilck J, Cannie M, Robrechts C, de Ravel T, Dymarkowski S, Van den Bosch T, Van Schoubroeck D: First trimester diagnosis of sirenomelia. Prenat Diagn 2006;26:684-8.

13 Patel S, Suchet I: The role of color and power doppler ultrasound in the prenatal diagnosis of sirenomelia. Ultrasound Obstet Gynecol 2004;24:684-91.

14 Sepulveda W, Corral E, Sanchez J, Carstens E, Schnapp C: Sirenomelia sequence versus renal agenesis: Prenatal differentiation with power doppler ultrasound. Ultrasound Obstet Gynecol 1998;11:445-9.