

**CASE REPORT**

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**Prenatal diagnosis of sirenomelia in the first trimester****Ahter Tanay Tayyar<sup>1</sup>, Ahmet Tayyar<sup>2</sup>, Mehmet Baki Senturk<sup>1</sup>, Ayse Disli Gurler<sup>3</sup>, Hulya Akgun<sup>4</sup>, Mehmet Tayyar<sup>3</sup>**<sup>1</sup>Department of Obstetrics and Gynecology, Zeynep Kamil Maternity and Childrens Training and Research Hospital, İstanbul, Turkey<sup>2</sup>Private Doctor, Kayseri, Turkey<sup>3</sup>Department of Obstetrics and Gynecology, Erciyes University Faculty of Medicine, Kayseri, Turkey<sup>4</sup>Department of Pathology, Erciyes University Faculty of Medicine, Kayseri, Turkey

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**Abstract**

Sirenomelia is a rare and fatal congenital anomaly. Early prenatal diagnosis by first trimester scan should be the aim to minimize the physical and psychiatric trauma related to the termination of pregnancy at advanced gestation. Nulliparous woman aged 23 years was referred for evaluation because of absence of legs at 12 weeks 6 days of gestation. The upper half of the fetus appeared normal. The lower extremities appeared to be fused together in fixed extension and mobility at the hip and knee joints was restricted. A single umbilical artery was demonstrated using color doppler. Also, omphalocele was detected and the final diagnosis was sirenomelia. Parents opted for the pregnancy termination due to the lethal prognosis for the fetus. Etiological factors of sirenomelia sequence are still unclear. Several heterogeneous risk factors have been reported, but all of them are debatable. Anomalies of kidneys, significant oligohydramnios and concomitant hypoplasia of the lungs make a very unfavorable prognosis for sirenomelia. Ultrasonographic diagnosis of the sirenomelia in second trimester may be prevented by severe oligohydramnios, which is caused by renal agenesis or dysgenesis. Termination of the pregnancy is an option due to a lethal prognosis. It should be offered to the parents when diagnosis of the sirenomelia is made. First trimester diagnosis will give opportunity to the patient more safer and less traumatic termination procedure.

**Keywords :** Sirenomelia, prenatal diagnosis, first trimester**Introduction**

Sirenomelia, the Mermaid Syndrome is a rare and lethal congenital anomaly with an incidence of one in 60,000 to 100,000 pregnancies. Musculoskeletal disorders such as lower extremity fusion, urogenital and gastrointestinal system abnormalities could meet the criteria of this syndrome. Anomalies of kidneys, significant oligohydramnios and concomitant hypoplasia of the lungs make a very unfavorable prognosis for sirenomelia [1,2]. The concomitant anomalies, such as anorectal defects, defects of urinary tracts, anomalies of pelvis and external genitalias, have extremely poor prognosis and almost always lead to death or very significant disability. 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, which is caused by renal agenesis or dysgenesis. Most cases are diagnosed postnally [3,4]. Early prenatal diagnosis by first trimester scan should be the aim to minimize the

trauma related to the termination of pregnancy at advanced gestation. In addition, where possible, a second ultrasonographic scan should be performed 4-6 weeks after the initial 7-8 weeks scan so that gross structural anomalies are detected and termination of pregnancy be considered earlier. In the literature few first trimester diagnosis of sirenomelia were reported, therefore we reported this case to remind the importance of early diagnosis of sirenomelia [5].

**Case Report**

Nulliparous woman aged 23 years was referred for evaluation because of absence of legs at 12 weeks 6 days of gestation. An antenatal ultrasonographic scan revealed a single live fetus. Crown rump length was 56.6 mm and nuchal translucency was 2mm. The upper half of the fetus appeared normal with both upper extremities seen separately and moving normally. However, the lower extremities appeared to be fused together in fixed extension and mobility at the hip and knee joints was restricted [Figure 1]. A single umbilical artery was demonstrated using color. Also, omphalocele was detected. Sirenomelia has 7 types radiographically. Our case was consistent with type 6, which means that there was only one femur, and tibias and fibulas were absent. Parents opted for the pregnancy termination due to the lethal prognosis for the fetus [Figure 2]. Cytogenetic analysis of the removed material was done, revealing normal karyotype. Pathological investigation supported clinical diagnosis.

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**Figure 1.** 12 weeks 6 days gestation fetus with fused lower extremities



**Figure 2.** 12 weeks 6 days gestation fetus with sirenomelia

## Discussion

The names “sirenomelia” or “mermaid syndrome” originate from the physical similarity of the affected fetuses to the mythical creature, mermaid. Mermaid was a magic women with lower part of the body in the form of fish tail. Fish tail is resembled by the fusion of the

lower fetal limbs and partial or full fusion of the feet. Etiology of sirenomelia is unknown [1]. Etiological factors of sirenomelia sequence are still unclear. Several heterogeneous risk factors have been reported, but all of them are debatable. Chromosomal syndromes, monogenic conditions and familial recurrence were not identified in any cases. But as a part of the caudal regression syndrome familial cases were reported. Maternal risk factors associated with sirenomelia are diabetes mellitus and younger maternal age. Maternal diabetes is reported in 2% of cases [6]. Orioli et al [7] observed that younger maternal age, less than 20, has a higher prevalence of sirenomelia. From 10 to 15 percent of sirenomelia sequence cases are in twin births, most of them monozygous, and only one fetus is affected. There is no clear environmental factor associated with sirenomelia sequence. Fetal exposure to Experimental animal studies showed that retinoic acid, cadmium, lead, Ochratoxin A, induced sirenomelia [8-10]. But Holmes [11] stated that there is no teratogen associated with sirenomelia. As a result of these conflicting data, more extensive epidemiologic study is needed to identify etiology of this very rare congenital malformation. Two theories have been proposed to explain the etiopathogenesis of sirenomelia; the vascular steal hypothesis and the defective blastogenesis hypothesis. Normally, the umbilical cord contains two arteries that originate from the iliac arteries, which return blood to the placenta. In cases of sirenomelia, the umbilical artery is single and arises from the abdominal aorta. The abdominal aorta distal to this branch directly bifurcates into iliac iliac arteries without giving an origin to renal or inferior mesenteric artery branches. These vascular abnormalities lead to vitelline artery steal of the blood supply to the caudal end of embryo, which leads to sirenomelia and associated anomalies. At blastogenesis, damage to the caudal mesoderm of the embryo between day 13 and day 22 of life results in merging, malrotation, and dysgenesis of the lower extremities [12]. Anomalies of kidneys, significant oligohydramnios and concomitant hypoplasia of the lungs make a very unfavorable prognosis for sirenomelia. The concomitant anomalies, such as anorectal defects, defects of urinary tracts, anomalies of pelvis and external genitalias, have extremely poor prognosis and almost always lead to death or very significant disability. While performing differential diagnosis one should exclude, caudal regression syndrome, Potter syndrome, Meckel-Gruber syndrome, renal dysplasia, variants of obstructive uropathy, severe intrauterine growth retardation syndrome that is extremely difficult to do in second or third trimester because of severe oligohydramnios. Recurrence risk of sirenomelia is sporadic and fetal karyotype is usually unremarkable [5,13].

## Conclusion

Prenatal diagnosis of sirenomelia may be difficult in the second or third trimester because of the severe oligohydramnios; it should be easier to diagnose sirenomelia in the first trimester. Sirenomelia is fatal in most cases due to the characteristic pulmonary

hypoplasia and renal agenesis. Termination of the pregnancy is an option due to a lethal prognosis. It should be offered to the parents when diagnosis of the sirenomelia is made. First trimester diagnosis will give opportunity to the patient more safer and less traumatic termination procedure.

### Author's contributions

This work was carried out in collaboration between all authors. Authors MT, ADG and HA managed the patient. Authors ATT, AT and MBS managed the literature searches, wrote the draft and final copy of the manuscript. All authors read and approved the final manuscript.

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