

DOI: <https://doi.org/10.56936/18290825-3.19v.2025-112>**SIRENOMELIA: A UNIQUE CONGENITAL ANOMALY  
(CLINICAL CASE)****MKRTCHYAN R.A.<sup>1</sup>, GHARDYAN G.K.<sup>1\*</sup>, ABRAHAMYAN L.R.<sup>1</sup>, KARALYAN N.YU.<sup>2</sup>,  
ABRAHAMYAN S.H.<sup>1</sup>, ABRAHAMYAN R.A.<sup>1</sup>**<sup>1</sup> Department of Obstetrics and Gynecology No. 2, Yerevan State Medical University after Mkhitar Heratsi, Yerevan, Armenia<sup>2</sup> Department of Pathological Anatomy and Clinical Morphology, Yerevan State Medical University after Mkhitar Heratsi, Yerevan, Armenia*Received 21.01.2025; Accepted for printing 15.05.2025***ABSTRACT**

Sirenomelia (Mermaid Syndrome) is a congenital malformation characterized by partial or complete fusion of the lower limbs and is often associated with pelvic bone anomalies, absence or underdevelopment of external genitalia, absence of the anus, and renal agenesis or dysgenesis. Although this syndrome is generally incompatible with life due to its frequent association with congenital visceral anomalies, there have been rare reported cases of newborns who have survived, though typically with a very short lifespan. Approximately 300 cases have been described in international medical literature, 15% of which are associated with twins, most often monozygotic. We present a sporadic case of sirenomelia in a 30-year-old Indian pregnant woman whose pregnancy was unsupervised (she did not attend prenatal consultations). In our case, premature labor began at 27 weeks of gestation. Ultrasound examination diagnosed intrauterine fetal death, malformation of the lower limbs, and oligohydramnios. After delivery, it became clear that we were dealing with a stillborn male fetus exhibiting features characteristic of sirenomelia. According to radiological examination results, all bony elements of the fetus's lower limbs were present. In studying the case, the theory of vascular steal was identified as a possible pathogenic basis. According to this theory, the changes comprising mermaid syndrome result from circulatory disturbances in the lower part of the fetus's body due to the presence of a single umbilical artery. To our knowledge, this is the first such case reported in Armenia. **Conclusion.** Sirenomelia is an extremely rare and lethal congenital defect. It becomes evident that diagnosis becomes increasingly difficult as gestation progresses, which underscores the importance of regular prenatal visits for the early detection of sirenomelia and other anomalies. An important component of diagnosis is also Doppler examination of umbilical vessels, since the presence of a single umbilical artery often underlies the pathogenesis.

**KEYWORDS:** *sirenomelia, mermaid syndrome, rare defect, single umbilical artery, oligohydramnios***CITE THIS ARTICLE AS:**

Mkrtchyan R.A., Ghardyan G.K., Abrahamyan L.R., Karalyan N.Yu., Abrahamyan S.H., Abrahamyan R.A. (2025). Sirenomelia: A Unique Congenital Anomaly (Clinical Case); The New Armenian Medical Journal, vol.19 (3), 112-116; <https://doi.org/10.56936/18290825-3.19v.2025-112>

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## INTRODUCTION

Sirenomelia, also known as mermaid syndrome, is the partial or complete fusion of the lower limbs. It is a rare and fatal congenital anomaly. The first case of sirenomelia was described in 1542 by Rocheus, and since then, approximately 300 cases have been reported [Thombare D et al., 2023]. Its incidence is 0.8–4 cases per 60,000–100,000 pregnancies. The etiology is not fully understood, but the following risk factors are identified: genetic predisposition, maternal diabetes mellitus, use of teratogenic drugs, hypoperfusion, cocaine use, landfill waters, and maternal age under 20 or over 40. It occurs more frequently in monozygotic, male fetuses and can appear in any ethnic group [Kucuk S, Kucuk IG, 2020; Tilahun T, Desta D, 2021; Tamene A, Molla M, 2022]. In most cases, no clear cause is found (sporadic), suggesting the influence of environmental factors or the presence of mutations. Most likely, sirenomelia is a multifactorial defect [Bösenberg A, 2005]. Sirenomelia is often associated with other anomalies: absence of external genitalia, rectal atresia, renal agenesis, absence of the urinary bladder, presence of a single umbilical artery, esophageal atresia, pulmonary hypoplasia, heart defects, omphalocele, diaphragmatic hernia, pelvic bone abnormalities, and meningomyelocele [Sriram P et al., 2010; Al Hadhoud F et al., 2017; Turgut H et al., 2017; Morales-Roselló J et al., 2022].

Sirenomelia can present with varying phenotypes — from the mildest form, where all bones of the fused limbs are present, to the most severe form, where no signs remain of the limbs having developed separately. Mermaid syndrome was classified into seven types by Stocker and Heifetz based on the presence of skeletal elements in the lower limbs. In Type I, the mildest form, all bones of the two fused limbs are present, and the fusion is limited to soft tissue. Type VII is the most severe form, where the malformed lower limb contains only one bone.

Two main hypotheses have been proposed regarding the pathogenesis: the vascular steal theory and the defective blastogenesis theory.

The vascular steal hypothesis is based on the

presence of a single umbilical artery of vitelline origin, which leads to impaired blood outflow to the placenta, resulting in poor blood supply to the lower part of the body.

The defective blastogenesis hypothesis suggests a primary defect in blastogenesis that occurs during the late stages of gastrulation, corresponding to the third week of pregnancy [Garrido-Allepuz C et al., 2011; Sahu L et al., 2013; Xu Tingting, 2018; Kavunga E.K. et al., 2019; Shojaee A et al., 2021].

Cases of sirenomelia often result in stillbirth or neonatal death within the first 1–2 days after birth. The high mortality rate is explained by the combination of lower limb defects with various visceral anomalies, especially renal defects. However, in rare cases, longer survival has been recorded due to the milder expression of visceral anomalies. Among those who survived, the longest living individual was Tiffany Yorks, who lived for about 27 years and successfully underwent surgery to separate the lower limbs before the age of one [Sathe PA, 2014; Samal SK, Rathod S, 2015; Kattel P, 2018].

## CASE REPORT

A 30-year-old pregnant resident of Yerevan city presented to the Republican Institute of Reproductive Health, Perinatology, Obstetrics, and Gynecology, complaining of cramping pain in the lower abdominal area and bloody discharge from the vagina. In her medical history, she does not report pregnancy; the current pregnancy is her first, at 27 weeks, and has been uneventful. During the pregnancy, she did not attend prenatal consultations. She denies somatic and gynecological diseases or harmful habits in her history. On physical examination, the general condition of the pregnant woman was assessed as satisfactory.

Ultrasound examination diagnosed intrauterine fetal death (fetal heartbeats were absent), severe oligohydramnios, normally located placenta with premature partial abruption, and a retroplacental hematoma measuring 4.0 x 3.0 cm. The estimated fetal weight was 1150 g. A defect of the fetal lower limbs was visualized, but due to severe oligohydramnios, it was not possible to determine the type of defect.



**FIGURE 1.** Stillborn with mermaid syndrome. a)front b)back

Medical induction of labor was performed. The woman delivered vaginally a stillborn, premature male fetus weighing 950 g and measuring 33 cm, with multiple developmental anomalies (mermaid syndrome). According to the Apgar score, it was 0-0. External examination of the fetus revealed fusion of the lower limbs with two separate feet, each foot having five toes. On the lower part of the anterior abdominal wall of the fetus, there was a penis-like formation with a blind-ending opening. An absence of the anus was also detected (fig. 1). X-ray examination did not reveal absence of fetal bones; all skeletal elements of the lower limbs were present.

Internal examination of the fetus revealed bilateral agenesis of the kidneys and ureters, absence of the urinary bladder; the liver, lungs, and heart were normally developed without macroscopic changes. The small intestines were normally formed, and the rectum ended blindly, filled with meconium. Microscopic examination of internal organs and



**FIGURE 2.** Histological examination of the umbilical cord, x20 (Hematoxylin-eosin).

tissues showed hyperemia, hemorrhages, and significant degenerative changes. A single umbilical artery was present (fig. 2).

### DISCUSSION

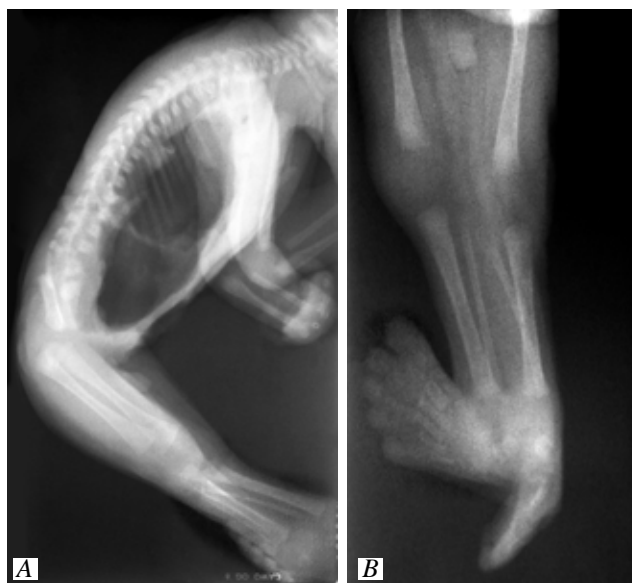
The exact etiology of sirenomelia is unknown. The influence of risk factors mentioned in international professional literature is absent in our case. In fact, this case, like most described cases, is sporadic [Bösenberg A 2005, Kucuk S, Kucuk IG, 2020, Tihahun T, Desta D 2021, Tamene A, Molla M 2022].

In this case, as in most cases described in international professional literature, sirenomelia was associated with visceral defects such as bilateral agenesis of the kidneys and ureters, absence of the bladder, underdeveloped penis-like formation with a blind-ending opening, blind-ending rectum, and absence of the anus [Sriram P et al., 2010, Al Hadhoud F et al., 2017, Turgut H et al., 2017, Morales-Roselló J et al., 2022].

Normally, there are two umbilical arteries. Considering the presence of only one umbilical artery in this case, the vascular steal theory is likely the basis for the malformation. In the absence of one umbilical artery, the aorta below the origin level of the remaining artery is significantly narrowed and deprived of many vessels supplying the kidneys, large intestine, and sexual organs. Therefore, a large volume of blood is forced through the existing single umbilical artery towards the placenta. As a result, disturbances occur in the blood circulation and nutrient supply to the lower limbs, leading to their developmental abnormalities [Garrido-Allepuz C et al., 2011, Sahu L et al., 2013, Xu Tingting, 2018, Kavunga E.K. et al., 2019, Shojaee A et al., 2021].

Ultrasound examination is the gold standard for diagnosing sirenomelia. Detecting sirenomelia by ultrasound is more likely during the first trimester of pregnancy when there is an adequate amount of amniotic fluid. Since the mermaid syndrome is often associated with renal anomalies, the detection of sirenomelia becomes difficult in later stages of pregnancy due to the reduction of amniotic fluid. Color Doppler examination is useful for diagnosing a single umbilical artery [Shojaee A et al., 2021]. In our case, the pregnant woman did not attend





**FIGURE 3.** Infantogram: all skeletal elements of the lower limb are present: **A)** skeleton, **B)** fused lower limbs

prenatal consultations, and no ultrasound examinations were performed during pregnancy, which is why the defect was not detected at early stages. The patient sought medical care after symptoms appeared, and ultrasound was performed only at 27 weeks, visualizing the lower limb defect; however,

due to severe oligohydramnios, it was not possible to determine the type of defect. The diagnosis of “sirenomelia” was made after delivery. Based on the performed X-ray examination conclusion, according to the Stocker and Heifetz classification, the fetus belongs to type I, meaning only the soft tissues of the two limbs are fused (fig. 3 a,b).

According to international professional literature data, sirenomelia mostly ends in stillbirth or neonatal death within the first postnatal days [Sathe PA, 2014, Kattel P, 2018, Samal SK, Rathod S, 2015]. In this case, intrauterine death was diagnosed.

### CONCLUSION

Sirenomelia is an extremely rare and lethal congenital defect. It becomes evident that diagnosis becomes increasingly difficult as gestation progresses, which underscores the importance of regular prenatal visits for the early detection of sirenomelia and other anomalies. An important component of diagnosis is also Doppler examination of umbilical vessels, since the presence of a single umbilical artery often underlies the pathogenesis.

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*Our journal is registered in the databases of Scopus,  
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LLC Print in "Monoprint" LLC

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