Paediatrica Indonesiana

p-ISSN 0030-9311; e-ISSN 2338-476X; Vol.63, No.1(2023). p.45-50; DOI: 10.14238/pi63.1.2023.45-50

Case Report

# Sirenomelia or "mermaid syndrome": an extremely rare case in Indonesia

**Qodri Santosa[1], Setya Dian Kartika[2], Irwan Nuryadin[3], Alfi Muntafiah[4]**

Sirenomelia, also known as mermaid syndrome, is a very rare lethal congenital disorder with multisystem malformations, characterized by fusion of the lower limbs. Here we report a preterm neonate with fusion of the lower limbs. The baby was born by cesarean section to a 27-year-old primigravida mother at 35 weeks and 3 days' gestation. There was no maternal history of hypertension, heart disease, asthma, or diabetes mellitus. At birth, the infant did not cry, had weak tone, a heart rate (HR) of <60 beats per minute (BPM), an Apgar score of 1/2, and a birth weight of 2,300 grams. The infant had an imperforate anus and no urogenital openings or external genitalia. There was a small penis-like protrusion without an aperture, such as in cases of ambiguous genitalia. The two lower limbs were fused, with five toes on each foot. Despite neonatal resuscitation, the infant died within 75 minutes of birth. The child was diagnosed with sirenomelia. This case report aims to emphasize the importance of early prenatal diagnosis and education of the patient's family. **[Paediatr Indones. 2023;63:45-50; DOI: 10.14238/pi63.1.2023.45-50** ].

***Keywords:*** *case report; congenital anomaly; lethal; lower limb fusion; mermaid syndrome; sirenomelia*

Sirenomelia or mermaid syndrome is a congenital disorder that is generally lethal.[1] Only a small number of cases survive.[2] The condition is characterized by multisystemic congenital malformations with wide phenotypic variability. The multiple malformations likely occur during intrauterine organogenesis.[3] This congenital anomaly is clinically characterized by fusion of the lower limbs and the pathognomonic sign of a single umbilical artery, that distinguishes it from other caudal regression syndromes (CRS).[1] The main characteristic of this malformation may be partial or complete fusion of the lower limbs, so that it looks like a single lower limb, giving the appearance of a mermaid.[4-6]

The incidence of sirenomelia varies from 0.8 to 1 case per 100,000 births.[5,6] Cases have been reported in various ethnic groups worldwide.[3, 7] The condition was initially described by Rocheus and Palfyn.[1,5] Mermaid syndrome is more common in boys than girls (2.7:1) and has been reported in cases of monozygotic twins.[6] Up to 22% of cases were born to mothers with diabetes mellitus, with a relative risk of 1:200-250.[1]

Postnatal management of sirenomelia is complicated and expensive, and most cases are fatal. Therefore, early prenatal diagnosis is essential, although the results will raise further ethical issues. This case report aims to provide an understanding of the importance of early prenatal diagnosis to reduce the risk of maternal morbidity associated with delivery (if the family decides to terminate the pregnancy); on the other hand, abortion raises serious ethical

From the Department of Child Health[1] and Department of Obstetrics & Gynecology[2], Universitas Jenderal Soedirman Medical School/Margono Soekarjo Hospital/Ananda Hospital, Ananda Hospital[3], Department of Biochemistry, Universitas Jenderal Soedirman Medical School[4], Purwokerto, Central Java, Indonesia.

**Corresponding author:** Qodri Santosa. Department of Child Health, Universitas Jenderal Soedirman Medical School/Margono Soekarjo Hospital, Purwokerto. Jl. Gumbreg No. 1 Mersi Purwokerto 53112, Phone 085720200984, email: qodrisantosa@gmail.com.

Submitted July 26, 2020. Accepted March 6, 2023.

problems. Ideally, the obstetrician should educate the family on the condition as soon as prenatal diagnosis is established. If the condition is not known until birth, the pediatrician, or other relevant healthcare provider, should provide education regarding the prognosis of the baby.

## The case

A primigravida mother, aged 27 years with 35 weeks + 3 days' gestation, visited a private hospital at Purwokerto, Central Java, Indonesia, because she was in labor. She had not undergone routine antenatal care, but had visited a general practitioner on occasion. She underwent a pregnancy examination by an obstetrician at five weeks' gestation; at that time, ultrasound did not detect a gestational sac (GS). The mother had no history of hypertension, heart disease, asthma, or diabetes mellitus.

General physical examination upon admission for labor was within normal limits. Gynecologic examination revealed a single intrauterine live fetus with breech presentation. The fetal HR was 136 BPM. The fundal height of the uterus was 22 cm (below normal for gestational age), indicating the presence of intrauterine growth restriction (IUGR). Ultrasound was not performed. Uterine contractions were every 10 minutes with a duration of 20 seconds each, giving the impression of imminent premature labor. Maternal laboratory results showed a hemoglobin level of 11.5 g/dL, a leukocyte count of 14,000/$\mu$L, hematocrits of 37.3%, an erythrocyte count of 4.36 x 106/$\mu$L, a platelet count of 242,000 /$\mu$L, a blood glucose level of 81 mg/dL, and non-reactive HBsAg and anti-HIV. Urinalysis showed slightly turbid color, pH 7, specific gravity 1.015, protein 0.15 g/dL, leukocyte esterase 2+, leukocytes 125 cells/$\mu$L, negative nitrite, leukocyte sediment 5-11/high-power field (HPF), erythrocyte sediment 1-3/HPF, and bacteria +1.

The baby was delivered by cesarean section on the indication of breech presentation, IUGR, and parturition not progressing. The baby did not cry, had weak muscle tone, a heart rate of <60 BPM, and multiple congenital anomalies. The baby's right and left lower limbs were fused (**Figure 1**), with five toes on each foot (**Figure 2**). The anal canal (**Figure 3**), urogenital orifice, and external genitalia (scrotum or labia majora) were absent, despite the presence of a small phallic-like protrusion as ambiguous genitalia (**Figure 1**). The infant's Apgar scores in the first and fifth minutes were 1 and 2, respectively, and birth weight was 2,300 grams. Despite the administration of neonatal resuscitation, the infant died within 75 minutes of birth. We did not perform any laboratory, radiological, or autopsy examinations. The patient was diagnosed as a low-birth-weight, small-for-gestational-age, preterm neonate, severe asphyxia, and sirenomelia (mermaid syndrome).

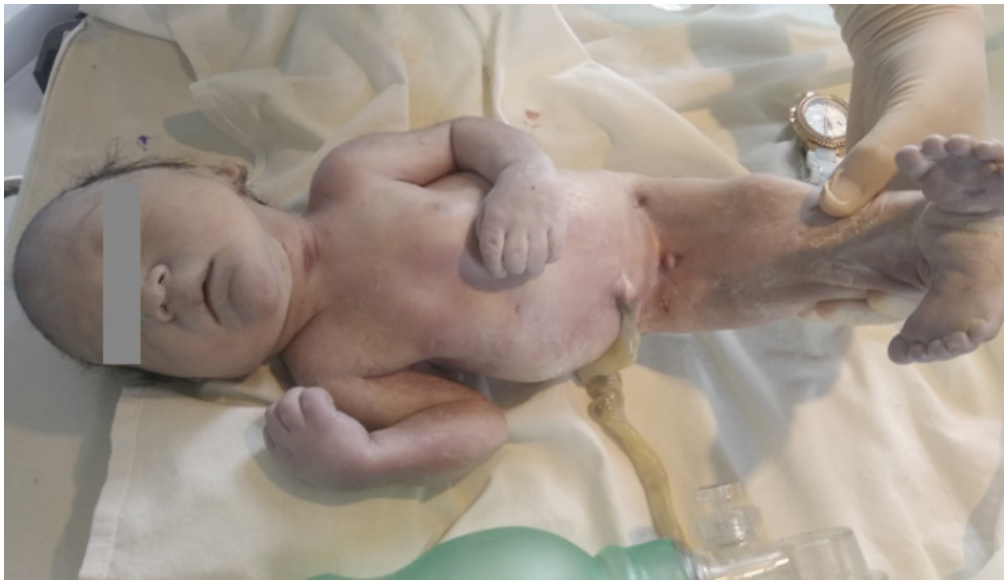

**Figure 1**. Fused lower limbs

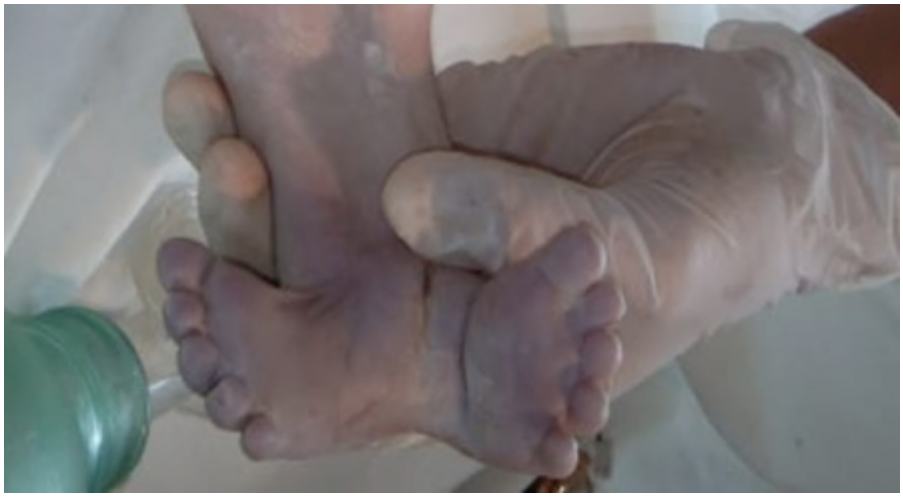

**Figure 2**. Fused lower limbs with five toes on each foot

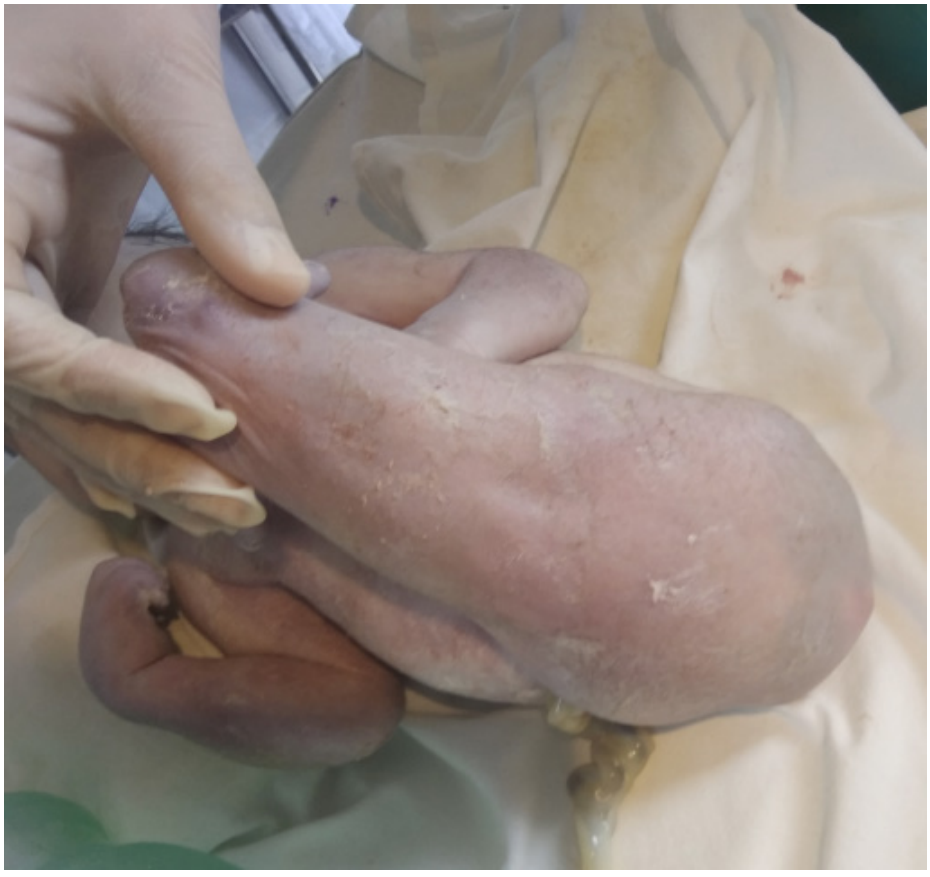

**Figure 3**. Fused lower limbs viewed from the back and imperforate anus

## Discussion

Sirenomelia is part of the CRS spectrum. CRS is characterized by sacrococcygeal or lumbosacrococcygeal agenesis, with a variety of features. These features can range from small anomalies in the lower vertebrae to more severe deformities, such as complete fusion of the lower limbs. The presence of complete or partial lower limb fusion in sirenomelia is called mermaid syndrome. Mermaid syndrome is the most severe form of CRS, and is usually accompanied by other severe fetal anomalies.[6,10,11]

To the best of our knowledge, this case is the third published case in Indonesia. Previously, Sutopo *et al.*[10] reported two cases of sirenomelia in 2017. Sporadic cases of mermaid syndrome have been reported in various countries in Europe, East, South, and Southeast Asia, the Middle East, and Africa.[1,2,4,6,7,9,11,13-23]

Caudal regression syndromes is a heterogeneous disorder with regards to etiology and pathogenesis. The development of CRS may involve genetic susceptibility and environmental factors.[10] Several theories been suggested.[6,11] The five pathogenetic theories for the development of sirenomelia include (1) embryologic insult; (2) vascular steal; (3) as part of CRS; (4) as part of VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies, and limb abnormalities); and/or (5) external forces on the lower extremities.

Some authors described that embryonic insult to the caudal mesoderm between days 28 and 32 of pregnancy causes vascular hypoperfusion (i.e., vascular steal), which might explain the limb fusion.[6,11,24] The embryonic insult theory states that nutrients are diverted from the tail end of the embryo, leading to poor growth/development, malformations, or even cessation of the development of organs or lower body tissue when the phase of development is still incomplete. However, this theory does not explain how abnormalities occur in the cranium, heart, or esophagus.6

Risk factors of mermaid syndrome include diabetes, teratogenic substances, and genetics. These factors might cause embryological insult through vascular stealing or primary defects in blastogenesis.[24] Uncontrolled maternal diabetes, especially in the first trimester, is a risk factor for sacral agenesis and caudal organ malformations, in general.[15,24] In our case, the mother did not have diabetes mellitus. The relationship between maternal diabetes and the incidence of sirenomelia has not yet been well-established. Only 3.7% of the cases reported were born to diabetic mothers,[11,17] hence, the etiological relationship between diabetes mellitus and sirenomelia is weak.17 However, several studies have shown that maternal uncontrolled diabetes is associated with CRS.[3,12,24,27,29]

Early prenatal investigations using ultrasound can detect structural anomalies in sirenomelia. This examination could reduce maternal morbidity by suggesting that a cesarean section be performed instead of normal delivery However, sirenomelia is so rare that it may not be detected in the first trimester without systematic screening.[13] Early prenatal diagnosis in the first trimester can be done using ultrasound at 4-6 weeks to 8-9 weeks of pregnancy.[4] Diagnosis of sacrococcygeal dysgenesis, an essential component of sirenomelia, can be done in the first nine weeks of gestation using transvaginal ultrasonography, although it is more frequently established in the second and third trimesters of pregnancy.[24,26] In 2001, Carbillon *et al.*[14] succeeded in detecting cases of sirenomelia at 13 weeks' gestation.

Ethical problems arise when multiple organ anomalies or gross structural anomalies are detected, as the parents are forced to decide between pregnancy termination or continuation. If the pregancy is allowed to continue, there is a risk of maternal trauma due to cesarean section, infant malformations, or even infant death, as in our case. The results of an early prenatal ultrasound can be used as a means of counseling mothers and their families. Some important considerations include gestational age at the time of diagnosis, severity of the malformation, the mother's decision, family beliefs, in addition to medical, cultural, and religious ethics.[11]

A case report stated that around 50% of sirenomelia cases were born at 8-9 months of gestation,[17] indicating that many cases are not detected in early pregnancy. In the present case, the mother underwent irregular antenatal care by a general practitioner and an ultrasound examination by an obstetrician at five weeks' gestation. No gestational sac was found at that time. From the initial antenatal

visit until birth, this case was not prenatally diagnosed.

The clinical characteristics of our case were complete fusion of the inferior extremities with five toes on each foot, imperforate anus, and absence of urogenital orifices. No external genitalia were seen, although there was a small penis-like protrusion without an orifice, appearing as ambiguous external genitalia. Based on these findings, our patient was diagnosed with sirenomelia (mermaid syndrome).

Several previous sirenomelia case reports reported malformations such as bilateral renal agenesis, complete or partial agenesis of the genitourinary system, and imperforate anus. The absence of external genitalia or ambiguous genitalia, a single umbilical artery, pulmonary hypoplasia, as well as gastrointestinal, vertebral, and heart anomalies have also been reported.[13,14,17]

A previous study reported on an infant with mermaid syndrome who was born prematurely at 25 weeks and died within the first 17 minutes. The child had malrotation and fusion of the entire lower legs with six toes and no external genitalia, urogenital openings, or anus. Autopsy revealed a single umbilical artery branching from the abdominal aorta, two polycystic kidneys, no distal part of the large intestine outside the caecum, rectal atresia, and reproductive organ agenesis.[9] Our patient did not undergo radiological examination or autopsy, so we have no knowledge on the details of the bone and visceral organ abnormalities.

Based on bone anatomy, sirenomelia is classified into seven types: type I with all thigh and leg bones present, type II with a single fibula, type III with absent fibula, type IV with partially fused femurs and fused fibulae, type V with partially fused femurs, type VI with a single femur, type VI with a single tibia, and type VII with a single femur and absent tibia. We could not determine the sirenomelia classification in our patient because no radiological investigation was performed. However, based on the results of physical examination (inspection and palpation), our case could be classified as type I, since both right and left feet had five toes.

Sirenomelia often includes fatal malformations, such as agenesis of the kidneys, as well as defects in the gastrointestinal system and other organs.[7] Multiple visceral anomalies[3,27] lead to mortality,[3,31], although there have been reported cases of extended life.[8,28]

Management of the baby with sirenomelia baby is time-consuming, complicated, expensive, and does not guarantee satisfactory results.[11,17] The primary therapeutic modalities involve surgical and medical management, especially surgical repair and maintaining systemic function.[11] Because of the expensive and challenging therapeutic modalities for sirenomelia, clinicians sometimes adopt conservative management as a therapeutic option, especially in patients with severe visceral abnormalities and poor prognosis.[26] We did not perform many diagnostic or therapeutic support modalities, except for neonatal resuscitation at birth.

The prognosis of the baby with sirenomelia is typically poor.[24] The severity of visceral organ anomalies is the most important prognosis. Sirenomelia patients with renal agenesis/dysgenesis,[6,17,26,27] and pulmonary hypoplasia17 have a dismal survival rate. Our sirenomelia patient did not cry at birth, had weak tone and a HR of <60 BPM, required resuscitation, and died about 75 minutes after birth. Other studies reported sirenomelia deaths at 15 minutes,[22] 20 minutes,[2,17] 30 minutes,[15] 12 hours,[15] and some at the age of 5 days after birth.[17,29] Some survivors of sirenomelia can live over the age of 5 years.

We have presented a fatal case of prenatally undiagnosed sirenomelia with multiple congenital anomalies. Early prenatal diagnosis is crucial to educate the family regarding management alternatives. Although early pregnancy termination reduces the risk to the mother during pregnancy and delivery, and the infant's chance of survival is small, the decision on whether to continue the pregnancy should be made in full consideration of medical ethics, prevailing laws, and the preferences of the mother and the family, including cultural and religious beliefs.

## Conflict of interest

None declared.

## References

1. Sikandar R, Munim S. Sirenomelia, the mermaid syndrome: case report and a brief review of literature. J Pak Med Assoc. 2009;59: 721-3. PMID: 19813692.

2. Kanagagiri, Suryaprakash, Singh YT, Singh KS. Sirenomelia apus – a case report. J Med Sci Clin Res. 2017;5:24993-6. DOI: https://doi.org/10.18535/jmscr/v5i7.113.
3. Garrido-Allepuz C, Haro E, González-Lamuño D, Martínez-Frías ML, Bertocchini F, Ros MA. A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations. Dis Model Mech. 2011;4:289-99. DOI: https://doi.org/10.1242/dmm.007732.
4. Valenzano M, Paoletti R, Rossi A, Farinini D, Garlaschi G, Fulcheri E. Sirenomelia. Pathological features, antenatal ultrasonographic clues, and a review of current embryogenic theories. Hum Reprod Update. 1999;5:82-6. DOI: https://doi.org/10.1093/humupd/5.1.82.
5. Reddy KR, Srinivas S, Kumar S, Reddy S, Prasad H, Irfan GM. Sirenomelia: a rare presentation. J Neonatal Surg. 2012;1:7.
6. Martinez-Frias ML, Garcia A, Bermejo E. Cyclopia and sirenomelia in a liveborn infant. J Med Genet. 1998;35:263-4. PMID: 26023366.
7. Pal S, Biswas R, Danda D, Chattopadhyay JC, Danda TK. Sirenomelia: mermaid syndrome. Int J Anat Res. 2014;2:689-91. DOI: https://doi.org/10.16965/ijar.2014.526.
8. Semba K, Yamamura K. Etiology of caudal regression syndrome. Human Genet Embryol. 2013;3:107. DOI: https://doi.org/10.4172/2161-0436.1000107.
9. Al Hadhoud F, Kamal AH, Al Anjari A, Fe Diejomaoh M. Fusion of lower limbs with severe urogenital malformation in a newborn, a rare congenital clinical syndrome: case report. Int Med Case Rep J. 2017;10:313-7. DOI: https://doi.org/10.2147/IMCRJ.S139067.
10. Sutopo M and Uli T. "The mermaid syndrome" - kasus serial. Cermin Dunia Kedokteran. 2017; 44: 337-8. DOI: https://doi.org/10.55175/cdk.v44i5.799.
11. Arora G, Bagga GR, Nandu VV. A rare case of twin gestation with sirenomelia and a coexisting normal fetus. J South Asian Feder Obst Gynae. 2017;9:360-2. DOI: https://doi.org/10.5005/jp-journals-10006-1529.
12. Kumar Y, Gupta N, Hooda K, Sharma P, Sharma S, Kochar P, *et al.* Caudal regression syndrome: a case series of a rare congenital anomaly. Pol J Radiol. 2017;82:188-92. DOI: https://doi.org/10.12659/PJR.900971.
13. Contu R, Zoppi MA, Axiana C, Ibba RM, Monni G. First trimester diagnosis of sirenomelia by 2D and 3D ultrasound. Fetal Diagn Ther. 2009;26:41-4. DOI: https://doi.org/10.1159/000236359.
14. Lynch SA, Wright C. Sirenomelia, limb reduction defects, cardiovascular malformation, renal agenesis in an infant born to a diabetic mother. Clin Dysmorphol. 1997;6:75-80. PMID: 9018422
15. Stocker JT, Heifetz SA. Sirenomelia. A morphological study of 33 cases and review of the literature. Perspect Pediatr Pathol. 1987;10:7-50. PMID: 3588246.
16. Taori KB, Mitra K, Ghonga NP, Gandhi RO, Mammen T, Sahu J. Sirenomelia sequence (mermaid: report of three cases. Indian J Radiol Imaging. 2002;12:399-401.
17. Pinette MG, Hand M, Hunt RC, Blackstone J, Wax JR, Cartin A. Surviving sirenomelia. J Ultrasound Med. 2005;24:1555–9. DOI: https://doi.org/10.7863/jum.2005.24.11.1555.