

Delivery of a Sirenomelia Misdiagnosed as IUGR with Anamnios

Salma Tahri Jautei*, Khaoula Laaboub, Malainine Himine, Rim Laaboudi, Mounia Yousfi

Department of Gynecology-Obstetrics and High Risk Pregnancy, Maternity Souissi, University Hospital Center IBN SINA, University Mohammed 5, Rabat, Morocco
Email: *salma-tahri@hotmail.com

How to cite this paper: Tahri Jautei, S., Laaboub, K., Himine, M., Laaboudi, R. and Yousfi, M. (2025) Delivery of a Sirenomelia Misdiagnosed as IUGR with Anamnios. *Open Journal of Obstetrics and Gynecology*, 15, 235-239.
<https://doi.org/10.4236/ojog.2025.152020>

Received: January 5, 2025

Accepted: February 22, 2025

Published: February 25, 2025

Copyright © 2025 by author(s) and Scientific Research Publishing Inc. This work is licensed under the Creative Commons Attribution International License (CC BY 4.0).

<http://creativecommons.org/licenses/by/4.0/>



Open Access

Abstract

Sirenomelia is a rare and lethal condition and its etiopathogenesis is unclear. The diagnosis is based on first trimester ultrasound showing a complete fusion of soft tissue in both lower limbs and other visceral abnormalities. We report in this article, the misdiagnose of sirenomelia as IUGR leading the other to continue the pregnancy until 35 weeks of gestation.

Keywords

Sirenomelia, Caudal Regression Syndrome, Early Diagnosis

1. Introduction

Sirenomelia is a rare malformation. It is caused by a primary defect of the caudal axial skeleton and damage to the primary streak, which appears due to a vascular steal phenomenon. It was first described in 1542 and compared to mermaid because of its characteristic appearance: a complete fusion of soft tissue in which both lower limbs are contained in a single skin sheath. This fusion may be partial or total. Sirenomelia appears with an incidence of 1 per 600,000 births. Diagnosis of sirenomelia in antenatal period by ultrasound in the first trimester is primordial so an interruption of the pregnancy can be proposed. A risk for sirenomelia can be also found in patients with poorly controlled diabetes mellitus [1] [2]. It's a lethal condition due mostly to associated visceral abnormalities [3]. In this article, we report a case of Sirenomelia diagnosed after emergency delivery.

2. Case Report

A 20 years old primigravida mother was referred at 35 weeks of gestation to the Department of Gynecology and Obstetrics of the University Hospital Souissi

(Rabat, Morocco), with a suspect Intrauterine growth restriction (IUGR) associated to anamnios. She hadn't any pregnancy follow-up and no antenatal ultrasound. Her past medical records showed type 1 diabetes for 12 years with insulin protocol. Her glycemic control during the pregnancy and before was not satisfying and her glycated hemoglobin at 6 weeks of gestation was at 8%.

She was admitted into the hospital for early labor with 3 cm dilatation and closer contractions. An ultrasound confirmed the anamnios with an estimated weight at 1800 g and a transversal presentation but no further abnormalities. She was then admitted to the OR for an emergency c-section and the delivery of a newborn with Apgar score 02 then died at 15 minutes of extra-embryonic life.

The examination at birth (**Figure 1**) showed a single lower limb and a single rudimentary foot. In addition, there was an esophageal atresia, an anal atresia and an unidentifiable external genitalia. Also, the examination (**Figure 2**) of the umbilical cord showed 1 umbilical artery and 1 umbilical vein. A radiography (**Figure 3**) was performed in post-mortem showed that there was a femur and 2 long bones. Unfortunately, the parents refused an autopsy for further examination.



Figure 1. A single lower limb.



Figure 2. The umbilical cord with 1 umbilical artery and 1 umbilical vein.



Figure 3. Radiography in post-mortem.

3. Discussion

The prevalence of sirenomelia is estimated to be 1 per 60,000 births [4] [5] with a sex ratio of 2 boys affected for every girl [4]-[6]. This condition is typically accompanied by other abnormalities, including a single umbilical artery and malformations such as anal atresia, renal agenesis, urogenital and esophageal atresia [3]-[10]. During the second trimester, an oligohydramnios may occur due to these malformations, leading to pulmonary hypoplasia.

Sirenomelia is classified into seven types according to Stocker and Heifetz [11].

The etiopathogenesis of this condition remains unclear. There is no evidence of chromosomal aberrations in humans or familial recurrences [8]. However, two main theories have been proposed in the literature. The first, proposed by Stevenson *et al.* in 1986, is the vascular steal theory [7] [8], which suggests that the presence of a single umbilical artery redirects blood flow away from the caudal mesoderm of the embryo during early embryogenesis (between 13th and 22th days of gestation), resulting in agenesis of midline structures and fusion of the lower extremities. The second theory involves defective blastogenesis during the third week of gestation, leading to caudal regression syndrome and interfering with the development of caudal mesoderm [3] [7]-[9].

In addition, certain risk factors may contribute to the development of Sirenomelia, including maternal diabetes and exposure to teratogenic agents during pregnancy, such as Retinoic acid, vitamin A, certain drugs, and cocaine [3].

Diagnosis of sirenomelia during the antenatal period can be challenging in the second or third trimester due to oligohydramnios. However, diagnosis is more feasible during the first trimester using transvaginal ultrasound, which can detect limb and visceral anomalies [12]. Furthermore, color and power Doppler imaging are critical for evaluating the vascular abnormalities [13].

Post-mortem autopsy remains a valuable method for studying fetal malformations. Some researchers have suggested imaging techniques, such as computed tomography (CT) and magnetic resonance imaging (MRI), may serve as alternatives to conventional autopsy, offering improved evaluation of internal organs

[14].

Misdiagnosis of sirenomelia can lead to the continuation of pregnancy, giving families false hope for fetal survival and potentially depriving them of the option to terminate the pregnancy during its early stages.

Healthcare professionals must prioritize proper care for pregnant women, particularly in populations with limited access to regular prenatal monitoring. Emphasis should also be placed on the critical importance of first-trimester morphological ultrasound in identifying anomalies.

4. Conclusion

Sirenomelia is a rare condition and the etiopathogenesis is still unclear. The diagnosis is made by an ultrasound in the first trimester to give the patient the choice of terminating the pregnancy at an early gestational age.

Consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

Funding

No funding or grant support.

Conflicts of Interest

The authors have no conflicts of interest relevant to this article.

References

- [1] Turgut, H., Ozdemir, R., Gokce, I., Karakurt, C. and Karadag, A. (2017) Sirenomelia Associated with Hypoplastic Left Heart in a Newborn. *Balkan Journal of Medical Genetics*, **20**, 91-94. <https://doi.org/10.1515/bjmg-2017-0001>
- [2] Rathod, S. and Samal, S. (2015) Sirenomelia: The Mermaid Syndrome: Report of Two Cases. *Journal of Natural Science, Biology and Medicine*, **6**, 264-266. <https://doi.org/10.4103/0976-9668.149227>
- [3] Opitz, J.M., Zanni, G., Reynolds, J.F. and Gilbert-Barnes, E. (2002) Defects of Blastogenesis. *American Journal of Medical Genetics*, **115**, 269-286. <https://doi.org/10.1002/ajmg.10983>
- [4] Kulkarni, M.L., Abdul Manaf, K.M., Prasannakumar, D.G. and Kulkarni, P.M. (2004) Sirenomelia with Radial Dysplasia. *The Indian Journal of Pediatrics*, **71**, 447-449. <https://doi.org/10.1007/bf02725639>
- [5] Houfflin, V., Subtil, D., Cosson, M., Valtille, E., Carpentier, F., Bourgeot, P., *et al.* (1996) Antenatal Diagnosis of 3 Caudal Regression Syndromes Associated with Maternal Diabetes. *Journal de gynécologie, obstétrique et biologie de la reproduction*, **25**, 389-395.
- [6] Agha, R.A., Franchi, T., Sohrab, C., Mathew, G., Kirwan, A., Thomas, A., *et al.* (2020) The SCARE 2020 Guideline: Updating Consensus Surgical Case Report (SCARE) Guidelines. *International Journal of Surgery*, **84**, 226-230.
- [7] Orioli, I.M., Amar, E., Arteaga-Vazquez, J., Bakker, M.K., Bianca, S., Botto, L.D., *et*

- al.* (2011) Sirenomelia: An Epidemiologic Study in a Large Dataset from the International Clearinghouse of Birth Defects Surveillance and Research, and Literature Review. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics*, **157**, 358-373. <https://doi.org/10.1002/ajmg.c.30324>
- [8] Stevenson, R.E., Jones, K.L., Phelan, M.C., Jones, M.C., Barr, M., Clericuzio, C., *et al.* (1986) Vascular Steal: The Pathogenetic Mechanism Producing Sirenomelia and Associated Defects of the Viscera and Soft Tissues. *Pediatrics*, **78**, 451-457. <https://doi.org/10.1542/peds.78.3.451>
- [9] Dueterhoeft, S.M., Ernst, L.M., Siebert, J.R. and Kapur, R.P. (2007) Five Cases of Caudal Regression with an Aberrant Abdominal Umbilical Artery: Further Support for a Caudal Regression-Sirenomelia Spectrum. *American Journal of Medical Genetics Part A*, **143**, 3175-3184. <https://doi.org/10.1002/ajmg.a.32028>
- [10] Al-Haggar, M., Yahia, S., Abdel Hadi, D., Grill, F. and Al Kaissi, A. (2010) Sirenomelia (*Symelia apus*) with Potter's Syndrome in Connection with Gestational Diabetes Mellitus: A Case Report and Literature Review. *African Health Sciences*, **10**, 395-399.
- [11] Islam, N., Mandal, B., Das, R.N., Bera, G., Mukherjee, S. and Chatterjee, U. (2017) Sirenomelia Associated with Discoid Adrenal and Lumbar Meningocele: An Autopsy Report. *Pathology—Research and Practice*, **213**, 1450-1453. <https://doi.org/10.1016/j.prp.2017.06.010>
- [12] Valenzano, M. (1999) Sirenomelia. Pathological Features, Antenatal Ultrasonographic Clues, and a Review of Current Embryogenic Theories. *Human Reproduction Update*, **5**, 82-86. <https://doi.org/10.1093/humupd/5.1.82>
- [13] Van Zalen-Sprock, M.M., Van Vugt, J.M.G., Van Der Harten, J.J. and Van Geijn, H.P. (1995) Early Second-Trimester Diagnosis of Sirenomelia. *Prenatal Diagnosis*, **15**, 171-177. <https://doi.org/10.1002/pd.1970150211>
- [14] Votino, C., Bessieres, B., Segers, V., *et al.* (2014) Minimally Invasive Fetal Autopsy Using Three-Dimensional Ultrasound: A Feasibility Study. *Ultrasound in Obstetrics & Gynecology*, **52**, 776-783.