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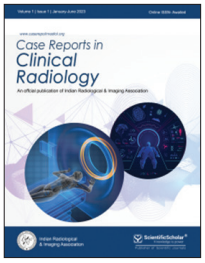

Case Report

Antenatal diagnosis of Sirenomelia, the Mermaid syndrome with bilateral renal agenesis

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Received: 26 October 2022
Accepted: 12 January 2023
EPub Ahead of Print: 13 March 2023
Published: 26 July 2023

DOI
10.25259/CRCR_25_2022

Quick Response Code:

ABSTRACT

A 26-year-old non-diabetic primigravida with 4 months of amenorrhea was appointed for a routine second-trimester anomaly scan. On inspection, the gravid abdominal girth of the lady did not correlate with her gestational age. Ultrasonographic examination showed a single live intrauterine gestation with anhydramnios. There was an absence of bilateral kidneys and urinary bladder with lying down adrenal sign suggestive of bilateral renal agenesis. The fetus also revealed caudal dysgenesis in the form of fusion of both lower limbs, seen as a single leg with 1 upturned foot and fused toes. There were no identifiable external genitalia and anus. The parents went ahead with medical termination of pregnancy after being informed about the condition and its incompatibility with life.

Keywords: Sirenomelia, Mermaid syndrome, Renal agenesis, Fused limbs

INTRODUCTION

Sirenomelia, also known as mermaid syndrome, is a rare fulminant multi-system congenital deformity. The incidence is reported to be one in 60,000–70,000 pregnancies. The "mermaid" in the name of this syndrome suggests that this condition is characterized by the fusion of lower limbs. There is a widely known association of Sirenomelia with severe urogenital and gastrointestinal malformations, making it a lethal disorder with an extremely low chance of live birth or survival post-birth. The live birth rates vary from 0.1:10,000 to 0.47:10,000 pregnancies based on various case reports. Early first-trimester diagnosis and induced abortion are the safest medical option.

CASE REPORT

A 26-year-old non-diabetic primigravida with 4 months of amenorrhea presented for a routine anomaly scan. The pregnancy was naturally conceived, with no history of usage of assisted reproductive technology. The patient has no history of diabetes mellitus (DM) or any other fetal anomalies in the mother's medical history. There is no significant family history. The marriage was non-consanguineous. The mother had no history of exposure to any intrinsic or extrinsic factors (such as teratogenic drug intake) associated with sirenomelia during her pregnancy. No previous obstetric ultrasound was available. The ultrasound revealed a single live intrauterine gestation, of average gestational age 19 weeks, 5 days with anhydramnios, absent bilateral kidneys, and urinary bladder. Limb examination on antenatal ultrasound revealed the presence of a single femur, with only two separate bones in the distal leg, instead of the usual four [Figure 1]. Upper limbs were grossly normal.

The humeral length was 30 mm corresponding to an average gestational age of 19 weeks, 5 days.

All cranial structures were visualized and normal. Nasal bone and lips were seen which were normal. No gross cardiac anomaly was seen. The chest circumference was decreased for the gestational age. The couple was counseled about the condition and its incompetency with life and medical termination of pregnancy was advised.

The pregnancy was terminated by induced conception. The abortus showed a single fused lower limb with an upturned foot and fused digits. There were no identifiable external genitalia and anus. The upper half of the body showed normal external development [Figure 2].

Radiographs of the abortus were obtained, which showed a single femur and two bones in the distal leg with morphology similar to that of the tibia. Both fibulas were absent [Figure 3]. This was consistent with Sirenomelia Type V as per Stocker and Heifetz classification. Ultrasound of the abortus confirmed bilateral renal agenesis with lying down adrenal sign and absent genitourinary tract [Figure 4].

Further, investigation could not be conducted because the parents refused an autopsy.

Few other conditions mimicking Sirenomelia include Amelia and Caudal regression syndrome. Amelia is complete agenesis of one or more limbs, while sirenomelia is characterized by fusion of both limbs, while caudal regression syndrome is a neural tube defect characterized by abnormal development of the caudal aspect of the vertebral column and the spinal cord. Limbs are not fused and there is often the presence of polyhydroamnios.

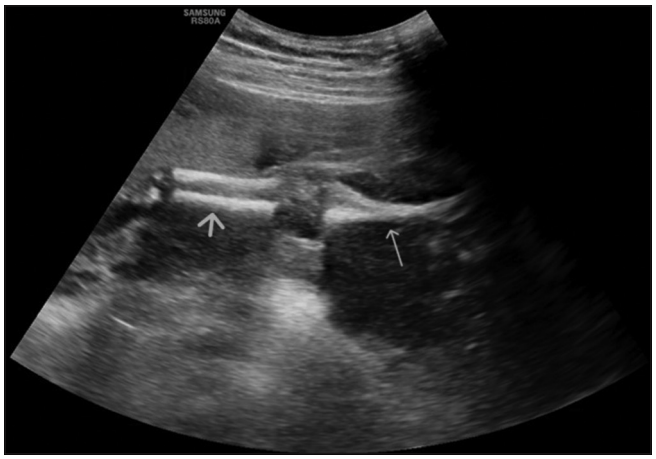

Figure 1: Antenatal US showing partly fused femur (long arrow) and only two distal bones (short arrow).

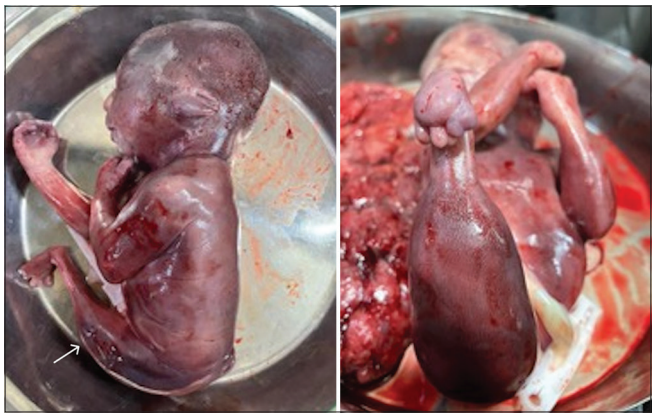

Figure 2: Images of the abortus with the arrow pointing at a single fused lower limb. The foot is upturned with fused digits. There is no identifiable external genitalia. Upper half of the body appears normal.

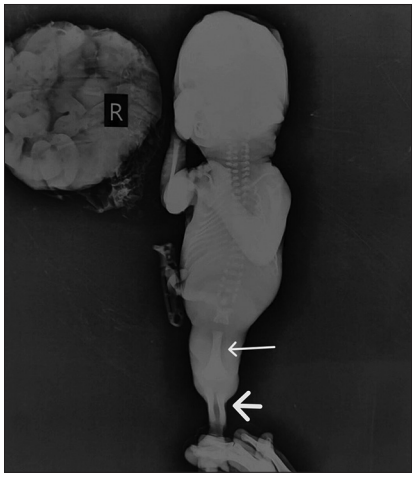

Figure 3: Radiograph of the abortus showing a single partly fused femur (long arrow) and two tibiae (short arrow). There is an absence of fibulae.

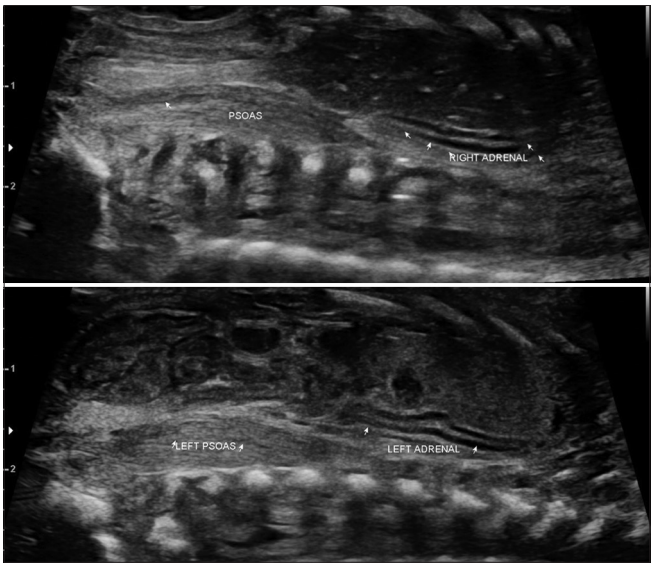

Figure 4: Ultrasound of the abortus confirming agenesis of both kidneys with lying down adrenal sign.

DISCUSSION

Sirenomelia or Mermaid syndrome is a rare and fatal congenital anomaly showing an incidence of 0.8–1 case/100,000 births. It shows male predilection with male-to-female ratio of 3:1. Till date, approximately 300 cases have been reported in the literature.[1]

Severe associated malformations of the gastrointestinal, genitourinary, cardiovascular, and musculoskeletal systems are usually associated with it, such as a single umbilical artery, imperforate anus, congenital cardiac anomalies, and abdominal wall defects. Oligohydramnios secondary to severe renal dysplasia is almost universally seen. However, in our case, we found anhydramnios with bilateral renal agenesis.

The exact etiopathogenesis of this malformation is unknown. Most of the cases are sporadic. Neonates born with mermaid syndrome usually show a normal karyotype.[2] Gestational DM is the only known maternal disease associated with the mermaid syndrome.[3-6] Nearly 22% of fetuses with this anomaly are found to have diabetic mothers.[1] Post-birth survival in cases of sirenomelia depends on the other associated visceral anomalies, predominantly the renal function, rather than the sirenomelia itself.

Congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India.[7] Early antenatal diagnosis is crucial to allow for timely intervention and to save the parents from the psychological mishap of delivering a malformed and incompatible with life fetus. Radiological modalities such as ultrasonography (US) and MRI play a crucial role in diagnosing antenatal fetal malformations. US is routinely performed during early pregnancy for dating, determination of the number of fetuses, assessment of early complications, and increasingly for evaluation of the fetus, including measurement of the thickness of the nuchal translucency. The second-trimester scan is a routine ultrasound examination, primarily used to assess fetal anatomy and detect any fetal anomalies. The majority of these studies are performed between 18 and 23 weeks.

In our case, a routine anomaly scan done at 19 weeks gestation showed anhydramnios suggesting an underlying genitourinary anomaly. Findings of fused femur and single lower limb were confirmed by radiographs of the abortus. The radiographic visualization of a single femur and two tibias helped in classifying the sirenomelia type according to Stocker and Heifetz classification. Bilateral renal agenesis was subsequently confirmed and documented by abortal US. A linear-shaped or "lying-down" adrenal gland is a sign often seen with the absence of the kidney in the renal fossa due to renal agenesis, renal ectopia, or horseshoe kidney. It is theorized that the presence of the kidney in the normal location within the renal fossa is important for the formation of the normal triangular inverted V or Y adrenal shape.[8]

Table 1: Stocker and Heifetz classification of sirenomelia.

Stocker/heifetz classification of sirenomelia	
Type I	All LL bones present- Two femur, tibiae, fibulae
Type II	Two femur & tibiae, medially fused fibula
Type III	Two femurs & tibiae, Absent fibula
Type IV	Partially fused femur, two tibiae, fused fibula
Type V	Partially fused femur, two tibiae, no fibula
Type VI	Completely fused femurs, single fused tibiae
Type VII	Completely fused femur, absent tibiae

Sirenomelia has been classified into several categories depending on the wide variety of limb malformation phenotypes. Commonly accepted classification is the Stocker and Heifetz method [Table 1]. It has seven types (I–VII) and is based on the presence or absence of the femur, tibia, and fibula.[9-12]

Recurrence risk is estimated up to 3–5%.[13] Hence, genetic counseling and antenatal folic acid tablets should be advised in upcoming pregnancies.

CONCLUSION

Sirenomelia is a rare but important fetal deformity that needs to be diagnosed early using the ultrasound. It is usually lethal when associated with geniotourinary or gastrointestinal malformations. Early ultrasound diagnosis is of immense value in these cases.

TEACHING POINTS

1. The pivotal role of antenatal ultrasound in timely diagnosis of fetal sirenomelia and deciding its classification.
2. Role of ultrasound imaging in diagnosing other anomalies associated with sirenomelia.
3. Importance of abortal radiographs in the classification of sirenomelia.

MCQs

1. Which of the type does fetus of sirenomelia with a partially fused femur, two tibias, and no fibula belong to as per Stocker Heifetz classification?
   a. Type V
   b. Type II
   c. Type III
   d. Type VI

Answer Key: a

2. Which of these associated anomalies should be looked for in the case of fetus with Sirenomelia and anhydramnios?
   a. Absence of adrenals

b. Absence of kidneys
c. Ectopia vesicae
d. Esophageal atresia

Answer Key: b

3. Which of the following feature is seen in Sirenomelia that helps to differentiate it from caudal regression syndrome?
   a. Polyhydroamnios
   b. Fusion of limbs
   c. Neural tube defect
   d. Compatibility with life.

Answer Key: b

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Reddy KR, Srinivas S, Kumar S, Reddy S, Prasad H, Irfan GM. Sirenomelia: A rare presentation. J Neonatal Surg 2012;1:7.
2. Orioli IM, Amar E, Arteaga-Vazquez J, Bakker MK, Bianca S, Botto LD, *et al*. Sirenomelia: An epidemiologic study in a large dataset from the International Clearinghouse of Birth Defects Surveillance and Research, and literature review. Am J Med Genet C Semin Med Genet 2011;157:358-73.
3. Al-Haggar M, Yahia S, Abdel-Hadi D, Grill F, Al Kaissi A. Sirenomelia (symelia apus) with Potter's syndrome in connection with gestational diabetes mellitus: A case report and literature review. Afr Health Sci 2010;10:395-9.
4. Chan BW, Chan KS, Koide T, Yeung SM, Leung MB, Copp AJ, *et al*. Maternal diabetes increases the risk of caudal regression caused by retinoic acid. Diabetes 2002;51:2811-6.
5. Taee N, Tarhani F, Safdary A. A case of sirenomelia in newborn of a diabetic mother. Int J Diabetes Metab 2005;5:83-7.
6. Taee N, Tarhani F, Goodarzi MF, Safdari M, Bajelan A. Mermaid syndrome: A case report of a rare congenital anomaly in full-term neonate with thumb deformity. Am J Perinatol Rep 2018;8:e328-31.
7. Patel SM, Vaishnav SB, Shah KN, Modi N, Raithatha NS, Phadke A, *et al*. Congenital malformation-a biological accident. Parity 2005;30:28-16.
8. Majmudar A, Cohen HL. "Lying-Down" adrenal sign: There are exceptions to the rule among fetuses and neonates. J Ultrasound Med 2017;36:2599-603.
9. Shojaee A, Ronnasian F, Behnam M, Salehi M. Sirenomelia: Two case reports. J Med Case Rep 2021;15:217.
10. Garrido-Allepuz C, Haro E, Gonzalez-Lamuno D, Martinez-Frias 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.
11. Torabizadeh Z, Naghshvar F, Nosrati A, Emadian O. Mermaid syndrome, Sirenomelia: A case report and review of literature. J Pediatr Rev 2013;1:56-61.
12. Boer LL, Morava E, Klein WM, Schepens-Franke AN, Oostra RJ. Sirenomelia: A multi-systemic polytopic field defect with ongoing controversies. Birth Defects Res 2017;109:791-804.
13. Fadhlaoui A, Khrouf M, Gaigi S, Zhioua F, Chaker A. The sirenomelia sequence: A case history. Clin Med Insights Case Rep 2010;3:41-9.

How to cite this article: Rani A, Sultan M. Antenatal diagnosis of sirenomelia, the mermaid syndrome with bilateral renal agenesis. Case Rep Clin Radiol 2023;1:92-5.