

The Sirenomelia sequence: a case report

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Sirenomelia or mermaid syndrome is a rare congenital condition with a wide phenotypic variation. It was originally named as caudal regression syndrome but it is now known that organs do not regress, they just do not form. An abnormal abdominal and umbilical vascular arrangement of affected individuals, and a primary abnormality in the generation of the mesoderm are two theories suggested for its genesis. Affected individuals show hypoplastic and fused lower limbs, vertebral abnormalities, and agenesis of the renal system, imperforated anus, and genital organs anomalies. Antenatal diagnosis in the first trimester is critical. We report a case of the mermaid syndrome associated with severe oligohydramnios, Potter's facies, unilateral renal agenesis, and absent external genitalia.

Keywords: lower limb fusion; mermaid syndrome; oligohydramnios; renal agenesis; sirenomelia

INTRODUCTION

Sirenomelia is a rare congenital condition with a prevalence of 1/100,000 pregnancies.¹ It is also referred to as mermaid syndrome because of the fused lower limbs which resembles the mermaid's tail. This feature is pathognomonic for the condition and the affected individuals have a variable range of vertebral abnormalities, anomalies of the genital organs, renal system, and imperforated anus.⁴

CASE

A baby born to a primigravida by spontaneous preterm vaginal delivery at home (at 33.5 weeks) was brought to the hospital with gasping respirations. The baby had a weak cry at birth. Despite of resuscitation, the baby survived for two hours only. Although autopsy was denied by the parents, antenatal ultrasonography had revealed a solitary cystic kidney with non visualization of left kidney and urinary bladder (possibly secondary to agenesis) along with severe oligohydramnios (amniotic fluid index of 2).

On examination, the baby had multiple visible anomalies (figure 1), which included Potter's facies with features like ocular hypertelorism, low-set ears and abnormal pinna, and flattening of the nose, urogenital anomalies like absence of urinary orifice, anal imperforation, absence of well differentiated external genitalia (an appendage resembling penis

was seen posteriorly but scrotal sacs or testes were absent) (figure 2), and limb anomalies i.e. a single inferior limb (resembling an ectopode of "mermaid") terminating in two slender limbs (stumps) (figure 3).

COMMENTS

The sirenomelia sequence, is a polymalformative syndrome which continues to cause many controversies concerning its etiopathogenesis. The name is derived from Greek mythology, in which there is a mention of creatures called, "sirens". These creatures had head and face of a female human being and the torso of an avian creature from the wings down. They were hazardous to sailors, whom they hypnotized with their captivating voices to later kill them. Over time, these creatures were depicted as more aquatic beings, and eventually with a full mermaid-like form.² This condition was first described by Kampmeier³ in 1927, and later a correlation between mermaid syndrome and visceral abnormalities was described in detail by Duhamel⁴ in 1961. Approximately 300 cases are reported worldwide of which 14 were from India.⁵ Sirenomelia is reported to be more frequent in the male sex, among monozygotic twins, and in babies born to mothers aged less than 20 years.^{1,3}

The exact etiology of Sirenomelia is still elusive, with some researchers trying to explain the precise cause, and others trying to find a genetic basis and mutations leading to this multisystem condition. This defect was previously thought to be the result of a wedge shaped early defect of the posterior axis of caudal blastema, allowing for fusion of primary limb buds at their fibular margins with absent or partial development of the intermediate caudal structures.⁶ However, Stevenson et al⁷ showed that Sirenomelia and its

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commonly associated defects are produced by an alteration in early vascular development. Rather than blood returning from the placenta via the usual paired umbilical arteries arising from the iliac vessels, blood returns to the placenta through a single large vessel, a derivative of the vitelline artery complex, which arises from the aorta just below the diaphragm. This vascular aberration leads to a “vitelline artery steal” through which blood flow and thus the nutrients are diverted from the caudal structures of the embryo to the placenta.⁷ It was initially thought to be a very severe form of caudal regression syndrome, but recent data suggests that both are pathogenically unrelated.⁶ Other theories have been proposed like the defective blastogenesis theory.⁸ Various possible genetic aberrations have also been associated with Sirenomelia sequence mouse models.⁹ Sirenomelia occurs in mice lacking *Cyp26a1*, an enzymic component that degrades retinoic acid, and mice that develop with reduced bone morphogenetic protein signalling at the caudal embryonic region. The phenotypes of these mutant mice indicate that the sirenomelia sequence in humans is associated with an excess of Retinoic acid signalling and a defective bone morphogenetic protein signalling in the caudal portion.⁹

FIGURE 1. Small for gestational age baby with Potters facies and fusion of lower limbs with absent external genitalia



Sirenomelia is also a condition with vast phenotypic variability. No two cases of Sirenomelia are similar. It is mainly characterized by lower limb abnormality, visceral malformations, vascular malformations, and spinal anomalies. It is basically a congenital defect of the lower torso characterized by the apparent fusion of the legs into one lower limb. This external phenotype is associated with a combination of lethal visceral abnormalities, most commonly urogenital and gastrointestinal.⁴ There have been

various attempts to classify the spectrum of limb phenotypes, which is not easy owing to the wide variations observed. Stocker and Heifetz⁸ classified Sirenomelia into type I to type VII, according to the presence of skeletal elements in the lower limb. Type I which is the mildest form, all bones in the fused limbs are present, and the fusion affects superficial tissues only. In type VII, the most severe form, only a single bone is present, with no definition of legs or feet.⁸ Malformations in the urinary tract are constantly associated with genital malformations. These mostly affect the external genitalia, which are either not present or represented by an unclear tag of tissue. Gastrointestinal anomalies commonly include a blind-ending colon, rectal atresia and imperforate anus.⁹ Among other abnormalities, lumbosacral and pelvic malformations, including sacral agenesis, malformed vertebrae and hemivertebrae, and corresponding anomalies of the central nervous system are commonly observed.⁸

FIGURE 2. Abnormal appendage on the dorsal aspect, which could be a malformed penis and imperforated anus.



Demonstrating the fused femur in the ultrasound clinches the antenatal diagnosis of this rare condition in addition to decreased distance between two femurs and decreased or absent mobility of the two limbs with respect to each other. Every attempt should be made to detect the condition in the first trimester

itself, since the oligohydramnios secondary to renal anomalies will obscure the picture in later gestation. Color Doppler ultrasonography will help to identify the single umbilical artery supplying the lower body of the fetus. In the second trimester, presence of severe oligohydramnios and renal agenesis should raise the suspicion of Sirenomelia.

The fused lower limbs resembling the “ectopode” of mermaid.



Ono T et al¹⁰ reported a case where they confirmed the diagnosis in the late second trimester with the help of a three dimensional computed tomography scan. Due to the lethal visceral abnormalities, Sirenomelia is incompatible with life with death occurring in the perinatal period.

CONCLUSIONS

This case of sirenomelia was not diagnosed in the first trimester, however severe oligohydramnios and renal agenesis were detected in the second trimester ultrasound. It had the classic phenotype with lower limb fusion, renal anomalies, ambiguous genitalia and sacral agenesis. Diagnosing this lethal condition antenatally is desirable so that possible termination of pregnancy can be offered at the earliest.

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Conflict of Interest:

No.

REFERENCES

- 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;157C(4):358-73. doi:10.1002/ajmg.c.30324. Epub 2011 Oct 14.
- Stahl A, Tourame P. From teratology to mythology: Ancient legends. *Arch Pediatr.* 2010; 17(12): 1716–24. doi: 10.1016/j.arcped.2010.09.004.
- Kampmeier OF. On sireniform monsters, with a consideration of the causation and predominance of the male sex among them. *Anat Rec.*1927; 34: 365.
- Duhamel B. From the mermaid to anal imperforation: the syndrome of caudal regression. *Arch Dis Child.* 1964; 36: 152-5.
- Samal SK, Rathod S. Sirenomelia: The mermaid syndrome: Report of two cases. *J Nat Sci Biol Med.* 2015 ; 6(1): 264–266.doi:10.4103/0976-9668.149227.
- Jones KL,ed. Smith's Recognizable Patterns of Human Malformation,7th ed. WB Saunders Company. Philadelphia,PA.2013:823.
- Stevenson RE, Jones KL, Phelan MC, Jones MC, Barr M, Clericuzio C et al. Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. *Pediatrics.* 1986; 78: 451-7.
- Stocker JT, Heifetz SA. Sirenomelia: morphological study of 33 cases and review of literature. *Perspect Pediatr Pathol.* 1987; 10: 7-50.
- Carlos G, Endika H, Domingo G, María LM, Federica B, Maria AR. A clinical and experimental overview of sirenomelia: insight into the mechanisms of congenital limb malformations. *Dis Model Mech.* 2011; 4(3): 289–99. doi:10.1242/dmm.007732.
- Ono T, Katsura D, Tsuji S, Yomo H, Ishiko A, Inoue T et al. Prenatal diagnosis of sirenomelia in the late second trimester with three-dimensional helical computed tomography. *Tohoku J Exp Med.* 2011;225(2):85-7.