

## Sirenomelia In A Non-Diabetic Mother– A Rare Case Report in India

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### Abstract:

**Background:** Sirenomelia is a rare birth defect in which two lower limbs fuse to form a single limb giving it an appearance of a mermaid's tail hence it is also termed as "mermaid syndrome". This peculiar morphotype is associated with anomalies involving multiple systems such as genitourinary and gastrointestinal tract. Although its cause is unknown it is believed to occur as a result of genetic and environmental factors.

**Case history:** We here discuss a case of a 23-year-old primigravida with 20 weeks 1-day gestation with a breech presentation with severe oligohydramnios and multiple cardiac defects. She was apparently diagnosed to have had a missed abortion at 8 weeks of gestation and was put on an unknown drug by a local doctor.

**Conclusion:** Sirenomelia with its bad prognosis calls for early detection of its diagnosis which can lead to timely medical intervention and save the patients a lot of mental and physical trauma.

**Key Word:** Sirenomelia, mermaid syndrome, sirenomelia sequence, sirenomelia syndrome, sirenomelisis

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### I. Introduction

Sirenomelia also synonymous with mermaid syndrome, sirenomelia sequence, sirenomelia syndrome, or sirenomelisis a rare, fatal congenital defect comprising of different degrees of lower limb fusion, thoracolumbar spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia<sup>[1]</sup>. The incidence of sirenomelia is 0.8-1 case/100,000 births<sup>[2]</sup>.

There are many controversies concerning its etiopathogenesis. Its classification as a variant of caudal regression syndrome (CRS) is questionable till date<sup>[3]</sup>. Its relationship with narrow pelvis syndrome (a narrowing of the pelvis with the ischions getting closer (to fusion), anal imperforation, renal agenesis or dysplasia, urinary tract dysplasia and anomalies of the external genital organs) and VATER (vertebral defect, anal atresia, interauricular communication; interventricular communication, tracheal and oesophageal atresia, and renal or radial agenesis) syndrome is debatable<sup>[4]</sup>.

A wide search of the literature revealed maternal diabetes as the most common cause of this condition followed by maternal exposure to drugs like cocaine, cyclophosphamide, tobacco, retinoic acid and heavy metal exposure.

We report this case as rare as this condition is probably secondary to maternal cardiac defects or due to unknown drug intake.

### II. Case history

A 23 year old primigravida with 20 weeks 1 day gestational age with breech presentation with severe oligohydramnios with known case of rheumatic heart disease (RHD) with mitral regurgitation (MR) with moderate pulmonary artery hypertension (PAH) with NYHA grade III dyspnoea and severe hyperthyroidism underwent termination of pregnancy. An interesting fact here is that, at 8 weeks of her gestation, the mother had consulted a local doctor who apparently diagnosed a missed abortion and had put her on an unknown drug. The baby aborted weighed 180 grams, with attached umbilical cord measuring 12cm and the placenta weighing 80gms.

The external examination revealed the following morphotype abnormalities: (Fig 1,2,3)

A single inferior limb

Undetermined sex (no external genital organs)

Potter's facies (ocular hypertelorism, low-set ears, receding chin and flattening of the nose)

Anal imperforation

Absence of a urinary orifice.

On autopsy the internal abnormalities found were: (Fig 4,5,6,7)

Umbilical cord cut section- two vessels  
Bilateral pulmonary hypoplasia  
Normal heart and cardiac vessels  
Normal stomach, liver, biliary ducts and pancreas  
Two plane-shaped suprarenal glands,  
Absent rectum  
A blind bowel  
Absence of renal arteries  
Single non-branching aorta.

The X-ray of the skeleton revealed: A single femur, sacral agenesis.(Fig 8)

**Diagnosis :Sirenomelia sequence grade VII<sup>[11]</sup>.**



Fig 1:Fetus showing a single lower limb, absence of urinary orifice and external genitalia



Fig 2: Fetus showing sacral agenesis and imperforate anus



Fig 3: Potter's facies: Face showing depressed nasal bridge, hypertelorism, low set and flattened ears, receding chin



Fig 4: Cut section of umbilical cord showing 2 vessels

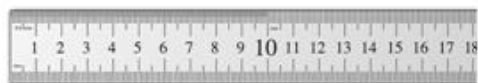


Fig 5: Pulmonary hypoplasia, normal heart, normal liver



Fig 6: Bilateral pulmonary hypoplasia



Fig 7: Renal agenesis and absence of rectum, single non branching aorta



Fig 8: Infantogram showing sacral agenesis, single femur, absent fibula and tibia

### III. Discussion

Sirenomelia is fatal in most cases because of the characteristic pulmonary hypoplasia and renal agenesis. About 50% of the children are born alive after eight or nine months of pregnancy<sup>[11]</sup>. The etiology is not completely known however many theories have emerged during the course of time.

#### Embryology:

At about 4 weeks of gestation, the embryological abnormalities related to medio-posterior mesodermic axis and caudal blastema is the basis of sirenomelia sequence<sup>[14]</sup>. At the fourth week of gestation, the cloaca is formed, kidneys are present at the pelvic level while the gonads are present intra-abdominally. So any injury to the caudal extremity of the embryo would, therefore, lead to abnormalities in the external and internal genital organs (though gonads are spared as they are intra-abdominal), kidneys, bladder, distal bowel and pelvic bones.

Prior to the proposition of the vascular steal theory<sup>[8]</sup>, the abnormalities involving the caudal extremities were categorized under Caudal Regression Syndrome (CRS) or Sirenomelia sequence, the spectrum

of which ranged from the mildest form of anal imperforation to sirenomelia which is a fusion of both the lower limbs<sup>[15]</sup>. Later on a new classification<sup>[14]</sup> has been adapted which thus distinguished between the two:

Caudal Regression Syndrome (CRS) :

It is associated with defects in the development of sacrum up till agenesia, spinal cord defects with urinary incontinence and abnormalities in the lower limb. Renal agenesia and anal imperforation are however not the constant features rendering this entity to be compatible with life.

Sirenomeliasequence :

This includes renal agenesia, absence of external and internal genital organs, imperforate anus with the absent rectum, sacral agenesia, dysgenesis of lumbar vertebrae and varying degrees of lower limb fusion.

Etiopathogenesis:

Maternal diabetes is the only maternal disease known to be associated with sirenomelia (2% of cases), although this is associated more frequently with Caudal Regression Syndrome (CRS)<sup>[5]</sup>.

Teratogenic drugs such as cocaine, organic solvents of fat and appetite suppressors (diethylpropion) have been implicated in some cases of CRS in humans<sup>[6]</sup>.

Several family cases of CRS have been reported, suggesting a possible genetic transmission. Welch et al<sup>[7]</sup> suggested an association between maternal diabetes and a genetic predisposition.

Vascular steal theory proposes that a mega-artery ensures the function of both umbilical arteries and diverts the blood flow from the embryo's caudal extremity toward the placenta, causing a nutritional deficit and a lack of development of the caudal extremity<sup>[8]</sup>.

Gardner et al<sup>[9]</sup> suggested "The pressure theory" which implicates that certain amniotic forces act on the caudal extremity of the embryo leading to its hypoplasia.

Dilorenzo et al<sup>[10]</sup> found that 9% to 15% of the cases of sirenomelia are associated with twin monozygotic pregnancies and that the relative risk is multiplied 100-fold in the case of twin pregnancies.

Classification of sirenomelia by Stocker and Heifetz<sup>[11]</sup>:

Type	Features
I	All thigh and leg bones are present
II	Single fibula
III	Absent fibula
IV	Partially fused femurs + fused fibulae
V	Partially fused femurs
VI	Single femur + single tibia
VII	Single femur + absent tibia

Sirenomelia is fatal in most cases because of the characteristic pulmonary hypoplasia and renal agenesia. Only about 50% of the children are born alive after eight or nine months of pregnancy<sup>[11]</sup>.

Murphy et al<sup>[12]</sup>, reported one case where a child born with sirenomelia survived; the infant was neurologically normal and had all features of sirenomelia sequence except for the presence of bilateral fused pelvic kidneys with renal dysplasia. Eventual separation of the lower extremities was performed.

In 1988 Tiffany Yorks underwent surgery to separate her legs before her first birthday. Though she still suffers mobility problems, at the age of 32, she is the oldest known surviving sufferer of the condition<sup>[13]</sup>.

This proves that the prognosis of sirenomelia is quite bad and the onus of a definite medical intervention lies on its early detection which can be as early as the first trimester, since oligohydramnios which is secondary to renal agenesia may pose difficulties in its diagnosis<sup>[16]</sup>.

#### IV. Conclusion

We thus report this rare case of sirenomelia. As in this case, the clinicians should be well aware of the other causes of this condition and give way for suspicion even in the absence of maternal diabetes and act promptly by getting appropriate antenatal investigations done to rule out any suspicion. In this 21st century where medicine has evolved with such advanced technology and when pregnancies are becoming more and more precious we believe that definite care and caution needs to be exercised to avoid anomalies as these and thus spare the parents and their offspring from the trauma that would result otherwise.

### References

- [1]. Reddy KR, Srinivas S, Kumar S, Reddy S, Prasad H, Irfan GM J Neonatal Surg. 2012 Jan-Mar; 1(1):7
- [2]. 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 Jan-Feb; 5(1):82-6
- [3]. Stevenson RE, Jones KL, Phelan MC, et al. Vascular steal: the pathogenic mechanism producing sirenomelia and associated defects of viscera and soft tissues. Pediatrics. 1986;78:451–7.
- [4]. Currarino G, Weinberg A. From small pelvic outlet syndrome to sirenomelia. PediatrPathol. 1991;11:195–210
- [5]. Valenzano M, Paoletti R, Rossi A, et al. Sirenomelia. Pathological features, antenatal ultrasonographic clues and a review of current embryogenic theories. Hum Reprod Update. 1999;5(1):82–6.
- [6]. Sarapong S, Headings V. Sirenomelia accompanying exposure of the embryo to cocaine. South Med J. 1992 May;85(5):545–7.
- [7]. Welch JP, Aterman K. The syndrome of caudal dysplasia: A review, including etiologic considerations and evidence of heterogeneity. PediatrPathol. 1984;2:313
- [8]. Adra A, Cordero D, Mejides A, et al. Caudal regression syndrome: Etiopathogenesis, prenatal diagnosis and perinatal management. ObstetGynecolSurv. 1994;49(7):508–76
- [9]. Gardner NJ, Breuer AC. Anomalies of heart, spleen, kidneys, gut and limbs may result from an overdistended neural tube. A hypothesis. Pediatrics. 1980;65:508.
- [10]. Dilorenzo M, Brandt ML, Veilleux A. Sirenomelia in an identical twin. A case report. J Pediatr Surg. 1991;26:1334
- [11]. Stockers JT, Heifetz SA. Sirenomelia : a morphological study of 33 cases and review of the literature. PerspectPediatrPathol. 1987;10:7–50.
- [12]. Murphy JJ, Fraser GC, Blair GK. Sirenomelia: case of a surviving mermaid. J Ped Surg. 1992 Oct 27;(10):1265–8
- [13]. <http://www.dailymail.co.uk/health/article-2767720/The-real-life-mermaids-Fatal-medical-condition-legs-fuse-womb-idea-mythical-creature-originated-says-expert.html#ixzz4GvazmnuU>
- [14]. Jones KL. Smith's Recognizable Pattern of Human Malformation. 4th edition. Philadelphia: Saunders; 1988. p. 574.
- [15]. Valenzano M, Paoletti R, Rossi A, et al. Sirenomelia. Pathological features, antenatal ultrasonographic clues and a review of current embryogenic theories. Hum Reprod Update. 1999;5(1):82–6.
- [16]. Ceylan et al. Sirenomelia, prenatal diagnosis Turk J ObstetGynecol 2016;13:50-2.

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