

Mermaids are Real!! Sirenomelia: A Case of Mermaid Baby

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ABSTRACT

We report a case of sirenomelia baby (mermaid syndrome) born to a thirty years old female at 36 weeks of gestation. It is a rare syndrome in which there is fusion of lower limbs.

Classification of caudal regression syndrome (CRS) from sirenomelia is still debated. According to some authors, this syndrome should be classified separately from caudal regression syndrome and is likely to be the result of an abnormality taking place during the fourth gestational week, causing developmental abnormalities in the lower extremities, pelvis, genitalia, urinary tract and digestive organs. Despite recent progress in pathology, the etiopathogenesis of sirenomelia is still debated.

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Introduction

Sirenomelia also known as “mermaid syndrome” is a polymalformative syndrome characterized by fusion of lower limbs, single umbilical artery, severe malformation of urogenital and lower gastrointestinal tract. It is a rare syndrome (0.8 to 4.2/100,000 births)^[1,2] which continues to cause many controversies concerning its etiopathogenesis.

Controversy exists in the literature regarding whether sirenomelia occurs as a separate entity or the extreme form of caudal regression syndrome (CRS). However, the presence of two umbilical arteries, non-lethal renal anomalies, non-fused lower limbs, abdominal wall defects, and abnormalities of tracheoesophageal tree, neural tube, and heart differentiate CRS from sirenomelia. In addition, CRS is strongly associated with maternal diabetes.^[3,4] The same applies regarding its relationship with narrow pelvis syndrome and VATER (vertebral defect, anal atresia, interauricular communication; interventricular communication, tracheal and esophageal atresia, and renal or radial agenesis) syndrome.^[5] We report a case of sirenomelia. We will discuss the classification and etiopathogenesis of this syndrome.

Case Report

A thirty years old female gave birth to baby by caesarian section performed at thirty six weeks of gestation for breech presentation and severe oligohydramnios. She was gravida 2 para2. Her husband was forty years old. She was not diabetic, non hypertensive, had no thyroid disorder or any evidence of infection and had no history of consumption of any teratogenic drugs. She had history of previous lower segment caesarian section with breech presentation and had delivered full term normal female child.

Antenatal ultra-sonography during this pregnancy revealed single viable fetus with breech presentation, oligohydramnios, and intra uterine growth retardation. Due to severe oligohydramnios the bladder, stomach, and spine could not be assessed. Kidneys were not visualized. The baby had respiratory distress but cried immediately after birth. Baby died within six hours of birth.

Autopsy revealed anomalous baby with weight 1.8 kilo grams. Body length was 40 cm, normal hair, large sized ears, nasal passage showed bleeding and eyes were closed.

The lower limbs were fused ending with two feet, right one with one big toe and left with two toes. (Fig1 A) There were no external genitalia or anus. (Fig1 B) There was a single umbilical artery. X-ray showed normal upper skeleton, two femurs, two tibias and one fibula (Fig 1 C).

Internal examination revealed normal 4 chambered heart. There was patent ductus arteriosus (Fig2D). Gastrointestinal



Fig. (1A): Showing fetus with fused lower limbs and large ear, (1B): Showing imperforated anus, (1C) : X ray showing two femur, two tibia and one fibula (white arrow)

tract (GIT) revealed imperforated anus, dilated rectum which was lying in pelvic cavity. (Fig 2 C) Rest of GIT was within normal limits with muconium in the cavity. One pancreas like structure was noticed. Two tiny cystic structures of size 0.2 x0.5 cm and 1x1 cm respectively suggestive of rudimentary kidneys were identified with attached thickened beaded cord like single structure (Fig 2 A, B). No bladder was identified.

Histopathology revealed congested lungs, spleen, liver, thymus and pancreas. Cystic structures revealed kidney with multiple cysts lined by cuboidal lining, primitive glomeruli, primitive tubules and intervening mesenchyme indicating cystic renal dysplasia (Fig 3A, B). Adrenals were identified which showed nodularity. Beaded cord like structures revealed tubules lined by ciliated columnar lining and small tubules lined by cuboidal lining indicating primitive testes and epididymis (Fig 3 C, D).

Diagnosis was offered as” Sirenomelia- Sympus Dipus type II” with imperforated anus, cystic renal dysplasia, absent urinary bladder, patent ductus arteriosus, primitive testicular tissue and nodular adrenals.^[6]

Discussion

Sirenomelia, best defined by Stevenson as “a limb anomaly in which the normally paired lower limbs are replaced by a single midline limb,”^[7] is an extremely rare congenital malformative disorder with a prevalence of approximately one per 100,000.^[8]

The syndrome of caudal regression was first used by Duhamel which consists of anomalies of the rectum, the urinary and genital systems, the lumbosacral spine, and the lower limbs. The most severe end of the spectrum is the fusion of the lower limbs and the major organ malformations, known as sirenomelia or mermaid syndrome, while the mildest end is imperforate anus.^[9] A debate still exists to whether CRS and sirenomelia

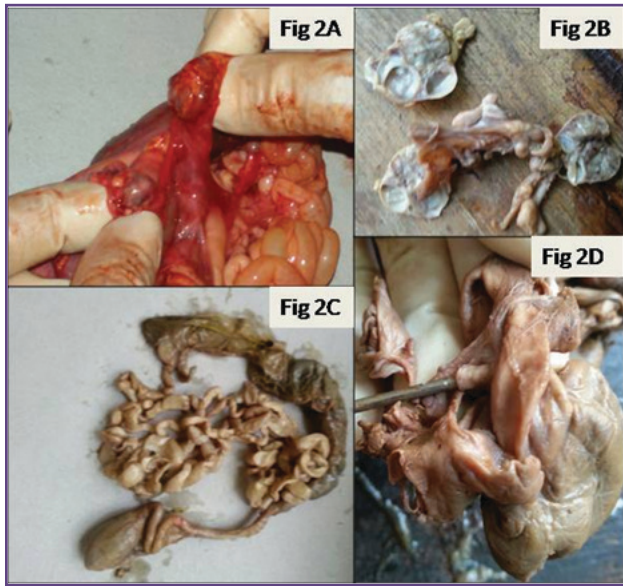


Fig. 2A: Showing rudimentary kidneys., **(2B):** Cut section of kidneys showing multiple cysts., **(2C):** Showing imperforated, dilated rectum., **(2D):** Showing patent ductus arteriosus

are separate entities or whether they represent parts of a pathogenic spectrum.

Duhamel endorsed a CRS-sirenomelia spectrum that encompasses variable severities of “an embryonal defect in the formation of the caudal region.” Whereas Jones segregated sirenomelia from CRS and asserted that they are unrelated pathogenetically. Apart from fusion of the lower extremities, a distinctive anatomic feature of sirenomelia is an aberrant umbilical artery that arises from the abdominal aorta, called “persistent vitelline artery.”^[10] A study by Orioli et al described the prevalence, associated malformations, and maternal characteristics among cases with sirenomelia.^[8]

Stocker and Heifetz classified sirenomelia in seven types:^[6]

- (I) All thigh and leg bones present.
- (II) Single fibula.
- (III) Absent fibulae.
- (IV) Partially fused femurs, fused fibulae.
- (V) Partially fused femurs, absent fibulae.
- (VI) Single femur, single tibia.
- (VII) Single femur absent tibiae.

Several etiologic factors have been suggested:

1. Maternal diabetes is the only maternal disease known to be associated with sirenomelia. However it is more common with CRS.

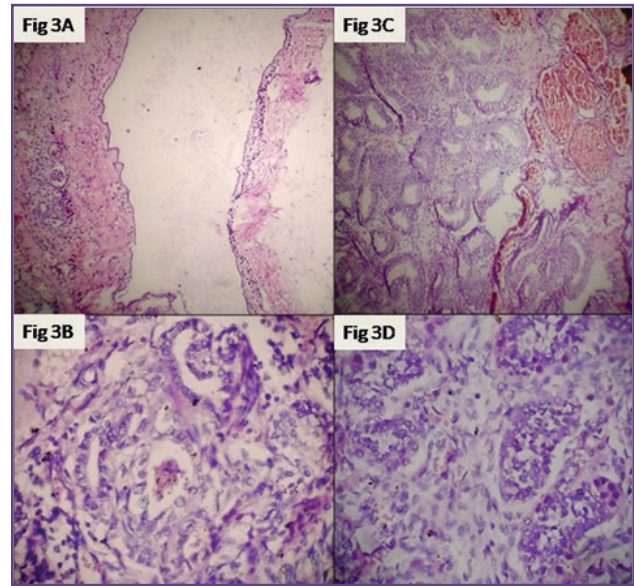


Fig. (3A): (10x, H and E) - Kidney showing cystic renal dysplasia., **(3B):** (40x, H and E) -Showing rudimentary glomerulus and tubules., **(3C):** (10x, H and E) - Showing epididymus., **(3D):** (40 x H and E) - Showing testicular tubules.

2. Teratogens like retinoic acid, cadmium, cyclophosphamide have reported to cause sirenomelia in mice. However no case of sirenomelia has been observed on accidental exposure of these products. Cocaine, organic solvents of fat and appetite suppressors (diethylpropion) has been implicated in some cases of CRS in humans.^[5]
3. Several family cases of CRS have been reported suggesting genetic predisposition
4. “Vascular steal” theory by Stevenson on the basis of dissection of the abdominal vasculature in eleven cases of sirenomelia has demonstrated a pattern of vascular abnormality.^[11] The common feature is the presence of a single large artery, arising from high in the abdominal cavity, which assumes the function of the umbilical arteries and diverts nutrients from the caudal end of the embryo distal to the level of its origin. The steal vessel derives from the vitelline artery complex, an early embryonic vascular network that supplies the yolk sac. Arteries below the level of this steal vessel are underdeveloped and tissues dependent upon them for nutrient supply fail to develop, are malformed, or arrest in some incomplete stage. In contrast to the prevailing view that sirenomelia arises by posterior fusion of the two developing lower limbs, these studies suggest that the single lower extremity in sirenomelia arises from failure of the lower limb bud field to be cleaved into two lateral masses by an

intervening allantois. Adra et al consider Stevenson's theory of the vascular steal as a possible etiopathogenic of CRS, while Stanton maintains that this theory cannot explain all the abnormalities encountered in the sirenomelia sequence and that the observations of Stevenson are a consequence and not a cause of the syndrome. ^[1,12]

5. 9% to 15% of the cases of sirenomelia are associated with twin monozygotic pregnancies. The relative risk is multiplied 100-fold in the case of twin pregnancies. ^[13]

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. Death occurs in the five days following birth. ^[6] Post-natal management requires the presence of kidneys, even if they are dysgenetic. ^[1] Murphy et al reported one case where a child born with sirenomelia survived. ^[14] Very few cases have been reported recently, one of them is by S. Samal et al who have reported two cases of sirenomelia where maternal drug abuse and overt diabetes may have been the cause of this rare anomaly. Both their babies died after few hours of birth. ^[15] Our case had single umbilical artery, fused lower limbs and cystic renal dysplasia. Baby died within six hours of birth.

Conclusion

Mermaids (Sirenomelia sequence) are real. It is a rare syndrome associated with various degree of vascular involvement in which prenatal diagnosis is essential for proper management. Probably study of molecular genetics will help to unravel the mystery.

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Competing Interests

None

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