SIRENOMELIA SEQUENCE –A CASE REPORT

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ABSTRACT

Sirenomelia sequence (Symelia) or "Mermaid syndrome" is a rare lethal congenital anomaly characterized by fusion, rotation, hypotrophy or atrophy of the lower limbs and severe urogenital abnormalities (usually absence of bladder and agenesis or dysgenesia of kidneys) leading to oligohydramnios. It is also always associated with agenesis or hypoplasia of diverse organs. It is a mermaid syndrome, a fact that is named so because the two fused lower limbs look like a mermaid's tail. The infants resemble the mythical siren. It occurs as a primary defect of blastogenesis affecting multiple midline primordia during the final stages of gastrulation at the caudal eminence. This leads to insufficient migration and differentiation of mesoderm which is responsible for the array of defects in the caudal region. This rare malformation is incompatible with life. These infants usually do not survive for more than 24 hours. Very few survive even after surgery. Only four known cases of children with the affliction are reported to be alive in the world today.

The aim of the study is to present a rare congenital malformation. Here we present such an interesting case of Sirenomelia. A 27 year old primi at 24 weeks of gestation was admitted to the hospital. Prenatal ultrasound demonstrated severe oligohydramnios and fusion of the lower limbs suggesting the diagnosis of Sirenomelia. Pregnancy was terminated and the fetus was submitted for detailed autopsy. The findings and review of the literature are presented in this report.

Key Words: Sirenomelia, teratogenesis, caudal regression, congenital malformations, vascular steal theory.

INTRODUCTION

Sirenomelia or mermaid syndrome represents an extreme form of caudal regression. It is a rare malformation sequence characterized by fusion of the lower limbs as well as involving multiple organ abnormalities[1]. The precise etiology of sirenomelia is not well understood. Some authors consider it as one of the clinical manifestations of the "caudal regression syndrome" where in developmental disruption of structures occur, derived from the caudal mesodermal axis of the embryo, and extended to various craniocaudal levels during the primitive streak stage. It is regarded as a part of the caudal dysplasia sequence. Etiologic theory is varied. There is no consistent data for the incidence of sirenomelia. It is estimated to be between 1.5-4.2 cases per 100,000 births [2] and reported as 0.8 in 100,000 births[3.] The condition occurs commonly in males, with a sex ratio of 2.7:1 [4]. The spectrum of lower limb anomalies ranges from simple fusion of soft tissues to the presence of single rudimentary limb. Sirenomelia is not associated with chromosomal aneuploidy.

The list of signs and symptoms for Sirenomelia includes the 9 symptoms:

- Fused legs.
- Absence of sacrum.
- Vertebral defects.
- Imperforate anus.
- Absence of rectum.
- Absence of external genitalia.
- Absence of internal genitalia.
- Renal agenesis.
- Absence of bladder.

Other associated anomalies are:

- Small ompholocele and Cord cyst [allantois or omphalomesenteric].
- Sirenomelia Sympodia [one fused foot].
- Umbilical artery arising from the superior mesenteric artery [normally arises from internal

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iliac artery]. Vessels in umbilical cord are one artery & one vein.

- ❖ Male fetus with absent penis & testes.
- ❖ Partial duplication of the small bowel and short colon.
- ❖ Potter's facies with low set mishapen ears.
- ❖ Lung hypoplasia.
- ❖ Bilateral poly cystic kidneys.

Thorough antenatal ultrasound scanning for fetal anomalies is necessary for prompt diagnosis of this rare anomaly. Later it is usually followed by therapeutic termination of pregnancy.

A CASE REPORT

A 27 year old female with 24- 25 weeks of gestation was admitted to the hospital and on ultra sound scan was diagnosed to have oligohydramnios and lack of fetal movements. The fetus demonstrated with fusion of lower limbs and sacral agenesis. At the level of kidneys, bilateral enlarged polycystic structure was identified and other multiple interesting anomalies were seen. The diagnosis of sirenomelia was made. Because of incompatibility with postnatal life, the pregnancy was terminated with the consent of the parents. Detailed fetal autopsy was done in anatomy dissection hall and the findings have been presented in the report. Since there were fused lower limbs seen, the fetus was subjected to radiological examination.

The affected fetus also presented the following gross features (Fig1&2). The eyes were amphibian like and the ears were floppy and low set.

Age corresponded to	:	24 – 25 weeks
Weight	:	450 grams.
Head circumference	:	16.8 cm.
Crown heel length	:	21.2 cm
Thorax circumference	:	17 cm.
Abdomen circumference	:	14.5 cm

The upper limb showed normal formation & measured about 10 cm in length. There was discoloration of right upper limb (Fig-1) . Lower limbs were fused & formed a single lower limb whose length was about 7.5 cm. The thighs and legs were fused with ill-defined muscles. The two feet were attached at the ankle joint and were rotated laterally, abducted and everted. The external genitalia were absent and it was represented as a small flap (Fig-1). There was an absence of anal orifice (imperforate anus) (Fig-2).

Internal features

Upon dissection the following features were observed: The placenta, brain, heart showed no remarkable findings. The right and left lungs were hypoplastic with no fissures (Fig-3).

The abdominal cavity showed multiple anomalies. The liver was small with agenesis of gallbladder and there was presence of annular pancreas around the duodenum. The stomach and spleen were normal (Fig-5). The intestines were malrotated with small intestine on the right and large intestine on left side. The blind end of the sigmoid colon opened into the cloaca (Fig-5) and there was an atresia of anorectal canal. Urogenital anomalies included (Fig-6) bilateral polycystic kidneys and two ureters opening into the cloaca. The bladder and urethra were absent. There was a small gonad on the left side of the abdomen. Cloaca received the two openings of the ureters and sigmoid colon. Vascular anomalies (Fig-4) seen were: abdominal aorta gave rise to superior mesenteric artery (SMA) which in turn gave rise to the steal vessel which continued as a single umbilical artery or aberrant umbilical artery (UMB.A) .Further there was caudal tapering of abdominal aorta as hypoplastic aorta and it ended at the pelvic brim. The inferior mesenteric artery (IMA) arose from abdominal aorta above the origin of superior mesenteric artery and was seen supplying only up to sigmoid colon. The aberrant femoral artery was seen rising from the hypoplastic aorta at the pelvic brim.

The Radiology findings were as follows (Fig-7)

Skull bones : Normal.

Upper Limb Bones : Normal.

Thorax and Abdomen: Ribs – 12 in no. present and all vertebrae were normal-(cervical, thoracic and lumbar) with Sacral agenesis. Lower Limb Bones : Hip Bones, Femora, Tibiae, Fibulae were present and were normal

Foot Skeleton : Tarsals, Metatarsals, phalanges

were present normally.

Impression : Soft tissue fusion in the region of thigh and leg up to the ankle region. Both feet separated and rotated laterally. It was diagnosed as mild mermaid syndrome, with sacral agenesis, restricted to skin and soft tissue.

DISCUSSION

Sirenomelia or the Mermaid syndrome is a rare congenital dysmorphic syndrome of the lower body segments with fusion of the lower limbs. The first reported case of mermaid syndrome was in the 16th century. It is characterized by fusion of lower limbs with sacral agenesis and other anomalies like imperforate anus, colonic atresia and rectal atresia, renal agenesis and absent bladder. Usually gonads are present and external genitalia absent.[1] Duhamel[5] in 1961 also mentioned all of visceral anomalies of the mermaid syndrome. He described imperforate anus, absence of genitalia (expect for gonads) and renal agenesis, gastrointestinal anomalies including vascular anomalies like aberrant single umbilical artery. This anomaly is a manifestation of caudal regression syndrome, which is a feature of the mermaid syndrome. The present case had all the above features mentioned by Lutz et al and Duhamel et al and except for renal agenesis, the fetus presented with polycystic kidneys and ureters opening in to cloaca. Cloaca also received the blind sigmoid colon (fig-6).

The spectrum of lower limb anomalies ranges from simple fusion of the soft tissues to the presence of single rudimentary limb. The syndrome has been classified into three types which is known as Sirtoris's classification[6].

SIRTORIS'S classification into 3 types according to the Number of feet.

1. Sympus Apus - absence of feet, only one tibia and one femur.
2. Sympus Unipus - presence of only one foot, two femora, two tibiae, two fibulae.
3. Sympus Dipus - presence of both feet and two fused legs. The fused appearance is of a flipper or fins.

Accordingly the present case belongs to the 3rd type with malrotated feet and associated with agenesis of gallbladder, annular pancreas, and intestinal malrotation with blind sigmoid colon ending in cloaca. The Kidneys were polycystic, ureters also ending in cloaca and agenesis of urinary bladder and urethra [3]. The index case also had vascular anomalies i.e. aberrant single umbilical artery arising from superior mesenteric artery and with hypoplastic aorta ending at the pelvic brim. It therefore may be representing a severe developmental field defect of the posterior axis of caudal blastema, resulting in fusion of the lower limb buds and also associated with agenesis or hypoplasia of viscera.[7]

Sirenomelia has been classified in a more detailed manner based on presence or absence of bones within the lower limb.[8]

Stocker's classification according to the fused bones:

Type – I : Paired femora, tibiae and fibulae.
Type – II : Paired femora & tibiae & single fused fibula.
Type – III : Paired femora &tibiae & absent fibula.
Type – IV : Partially fused femora &tibiae & single fibula.
Type – V : Partially fused femora & tibiae & absent fibulae.
Type – VI : A single femur & tibia with absent fibulae.
Type – VII: A single femur and absent tibiae and fibulae.

Our index case fitted into type I with sacral agenesis (x-ray fig -7) and with severe multiple internal anomalies and therefore incompatible with life. According to Barr M[9] defective intermediate mesoderm leads to abnormal kidneys. This case presented with polycystic kidneys with ureters opening into cloaca along with blind sigmoid colon, vascular anomalies, GIT anomalies and hypoplastic lungs. Three pathogenic theories have been proposed to explain this malformation. [10]

1. A pressure theory (oligohydramnios and intrauterine force acting on the caudal end of the embryo may impede normal rotation of limb buds).
2. Primary failure in the development of caudal somities that leads to defective development of lower parts of the embryo.
3. A lack of nutritional support of the caudal region of the embryo.

A “vascular steal” theory has been proposed to explain the various malformations of sirenomelia fetuses [10]. There's a diversion of the blood flow from caudal structures of the embryo to the placenta, and in consequence the caudal somites do not develop normally .The steal vessel derives from the vitelline artery complex that supplies the yolk sac i.e. superior mesenteric artery. Arteries below the level of this steal vessel are underdeveloped and the tissues arrest in some incomplete stage of development. Caudal regression and sirenomelia spectrum have been correlated with an aberrant single umbilical artery. The present case (Fig-4) had one umbilical artery continuing from superior mesenteric artery, which is a steal vessel. Abdominal aorta beyond the origin of superior mesenteric artery was hypoplastic and ended at pelvic brim. The pelvic organs were absent in the index case. The sigmoid colon and two ureters ended in a blind cloaca. There was complete absence of external genitalia with imperforate anus. It also presented with aberrant femoral artery from the hypoplastic aorta which explains fused lower extremities. Sirenomelia arises from failure of the lower limb bud field to be cleaved in to two lateral masses by an intervening allantois. This explains the vascular steal theory. [10]

Sirenomelia has also been described in association with maternal diabetes, cocaine or snuff exposure and monozygotic twins; the incidence being 150 times greater than in singletons.[12 &13] Absence of chromosomal abnormalities and familial inheritance has been noted in almost all cases. . The precise etiology of the present case of sirenomelia still remains unknown as mother was non diabetic and not exposed to any teratogenic agents.

Prognosis : Anomalies of kidneys, significant oligohydramnios and concomitant hypoplasia of the lungs make a very unfavorable prognosis for sirenomelia. Some milder forms of Sirenomelia with survival have been described in the literature. Usually these cases had only soft tissue fusion of the legs. But the concomitant anomalies, such as anorectal defects, defects of urinary tracts, anomalies of pelvis and external genitalia, have extremely poor prognosis and almost always lead to death.

CONCLUSION

The present case was a rare variant of mermaid syndrome of unknown etiology. It may be due to caudal dysplasia sequence associated with vascular steal vessel. Sirenomelia or mermaid syndrome represents a rare congenital malformation that is incompatible with life. Ultrasound may be useful in early antenatal detection of this anomaly. Survival is extremely rare due to multiple agenesis or hypoplastic viscera associated with fused lower limbs. In view of the bad prognosis earlier intra-uterine diagnosis allows less traumatic therapeutic abortion.

Mermaid syndrome may be a rare condition but still it is heart wrenching to hear about such cases. So let us end with a prayer that this condition becomes rarer than it is & every baby in this world is born healthy and happy. Amen.

Fig.1 Sirenomelia – Anterior View

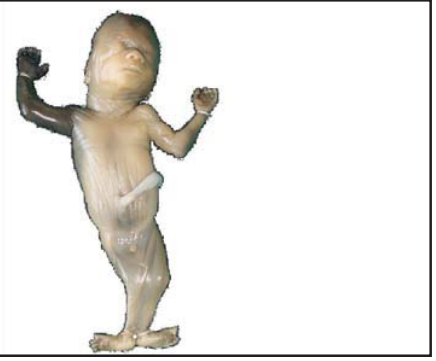


Fig.2 Sirenomelia – Posterior View

Fig.3 Dissected specimen with open thorax

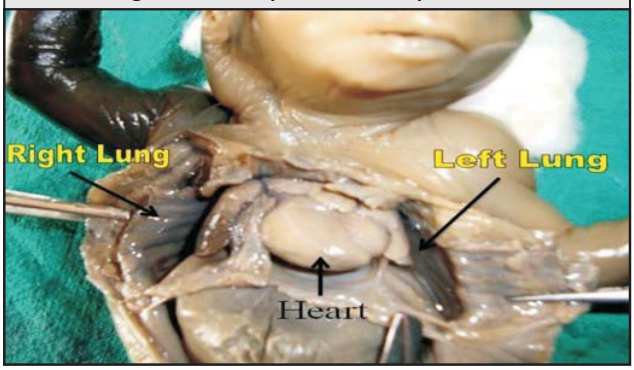


Fig.4 Dissected specimen with open abdomen showing Falciform ligament (Falciform L, Superior Mesenteric Artery (SMA), Inferior Mesenteric Artery (IMA), Aorta, Femoral Artery (Femoral.A), Umbilical Artery (UMB.A)

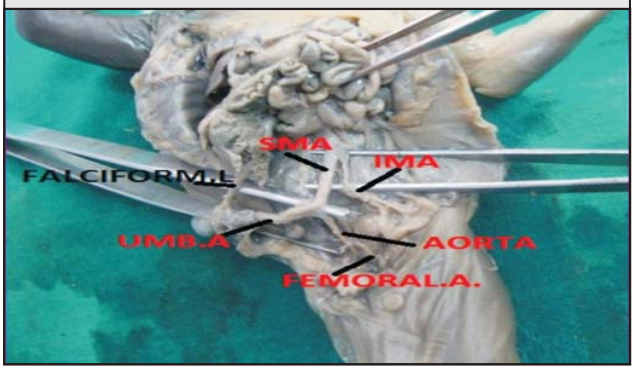


Fig.5 Dissected specimen with open abdomen showing abdominal organs

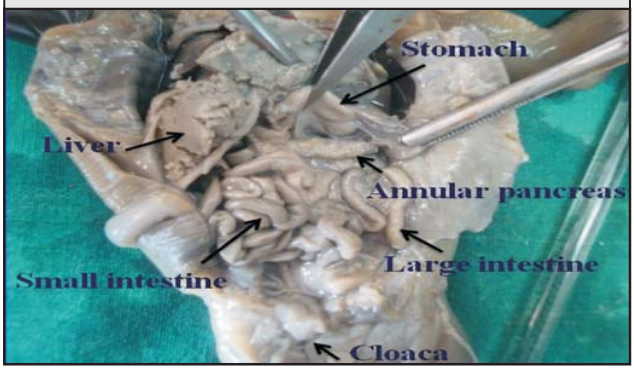


Fig.6 Dissected specimen with open abdomen showing abdominal organs

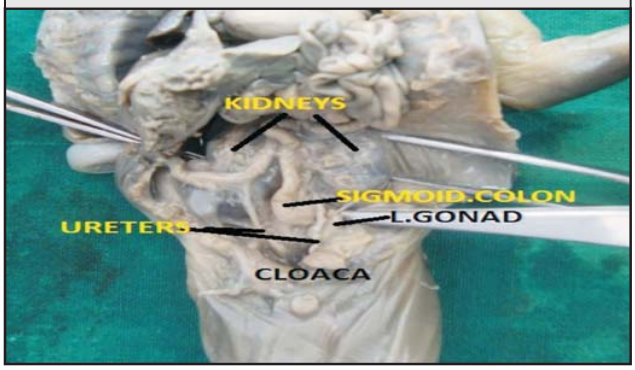
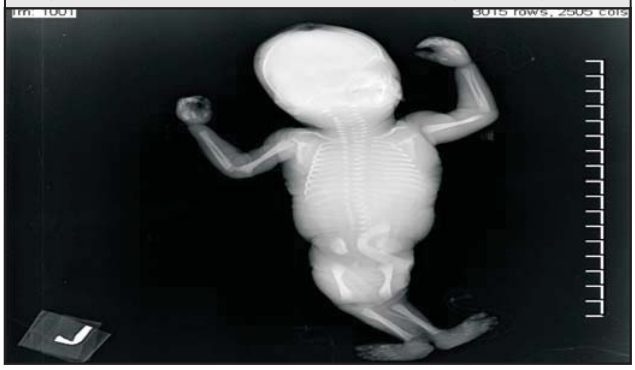


Fig.7 X-Ray of fetus with Sirenomelia showing sacral agenesis



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