

## Sirenomelia; the Mermaid Syndrome: A case report

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### Abstract

Sirenomelia, also known as "Mermaid Syndrome" is a very rare congenital anomaly with an estimated incidence of 1 in 100,000 pregnancies. It is usually fatal with isolated living patients reported. The characteristic feature of the Mermaid syndrome is complete fusion of the lower limbs (hence the name; mermaid). Other commonly associated features are renal agenesis, absent external genitalia and other gastrointestinal defects. It has also been associated with single umbilical artery, skeletal defects, and cardiac abnormalities. We hereby report a case of Sirenomelia or Mermaid syndrome, diagnosed after mid trimester medical termination of pregnancy ((20 weeks gestation) for ahydramnios with bilateral absence of kidneys. Only after abortion fetus was recognized to be having mermaid syndrome. Further, fetal autopsy was offered which revealed additional fetal anomalies in terms of absent kidneys, urinary bladder, and rectum. Sex of the abortus could be detected only after identification of one ovary as external genitalia and uterus were also absent.

**Keywords:** Sirenomelia; Mermaid syndrome; Medical termination of pregnancy

### Introduction

Sirenomelia is an extremely rare fetal anomaly. Primarily it is characterized by fused lower limbs, due to this reason it is also known as Mermaids Syndrome (as the typical mythological figure represents fused lower limbs) [1]. It was originally described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens. This rare fetal anomaly is usually fatal within few days of birth. More than half the cases of sirenomelia result in stillbirth. There has been association of maternal diabetes mellitus with Sirenomelia [2,3]. We hereby describe a case of Mermaid Syndrome diagnosed after the medical termination of pregnancy (MTP) was done for ahydramnios with bilateral absent kidneys.

### Case Report

A 27 year old, para 1, at 20 weeks of gestation was admitted for termination of pregnancy due to ultrasound (US) diagnosis of multiple congenital anomalies. Patient was booked and adequately supervised case of our institute. On routine malformation US scan she was diagnosed to have ahydramnios along with non-visualization of fetal kidneys. There was no personal or family history for diabetes mellitus, hypertension or congenital abnormality. Her previous pregnancy was also uneventful. There was no other risk factor for fetal malformation

i.e. maternal drug abuse, any therapeutic administration of teratogens or radiation therapy. The couple was counseled for MTP. Abortion was done using oral mifepristone 200mg followed 48hrs later by vaginal misoprostol 200µg every 4 hourly. The aborted fetus was having single lower limb and a foot with fused toes (Figure 1a). The sex of the fetus could not be ascertained externally as external genitalia were absent. The anal opening was also absent (Figure 1b). The fetus weighed 240gms and placental weight was 75gms. The examination of umbilical cord revealed a single umbilical artery and an umbilical vein.



**Figure 1:** a) Fetus showing sirenomelia, with fused lower limbs. B) Dorsal surface of fetus showing fused lower limbs similar to mermaid.

## Autopsy

The couple was advised fetal autopsy which revealed partially fused femurs and tibia, with absent fibula (Type III as per classification by Stocker & Heifetz et al.) [3]. There were eight toes in the single foot seen. Both the kidneys, urinary bladder and rectum were absent, small ovary was seen but there was no uterus. It is classified as symphysunipus (one foot present), as per classification by Forster et al.

## Discussion

Sirenomelia is an extremely rare & lethal congenital anomaly. Only isolated survivors have been reported till date. It is primarily associated with fusion of the lower extremities, bilateral renal agenesis, and absence of the sacrum, rectum, and bladder. Other anomalies associated with sirenomelia are heart and abdominal wall defects, skeletal abnormalities. Diagnosis is commonly made in the second trimester with oligohydramnios as the alerting sign. In our case, the diagnosis was primarily of anhydramnios with bilateral renal agenesis during routine US anomaly scan. Subsequently, the final diagnosis was confirmed on fetal autopsy. Additional abnormalities seen in our case were Potter facies (secondary to anhydramnios), single umbilical artery, imperforate anus and absence of external genitalia.

The exact cause of Sirenomelia is not known. Various hypotheses have been generated regarding its pathogenesis. Maternal diabetes has also been implicated in etio-pathogenesis. Various teratogenic substances have been implicated e.g. retinoic acid, cadmium, anti-epileptic drug (Lamotrigine), cyclophosphamide and cocaine abuse. Embryologically, it is a caudal blastemal defect, due to persistence of vitelline artery [4]. The diversion of blood from caudal portion of embryo via abdominal umbilical artery/ "vascular steal" has been proposed as the primary mechanism leading to Sirenomelia. Altered oxidative metabolism from maternal diabetes may cause increased production of free oxygen radicals in the developing embryo, which may be teratogenic [2-4].

Even though few authors have suggested first trimester US diagnosis of Sirenomelia earliest by the 9<sup>th</sup> week of gestation [5]. Nonetheless, it is prudent to mention that it is a very rare anomaly with incidence of 1 in 100,000 live births. A high index of suspicion is required for earlier diagnosis of this rare anomaly

especially in fetuses with absent or grossly reduced liquor. Ideally, women should be evaluated by detailed sonography in subsequent pregnancies too, though the incidence of recurrence of this anomaly is not exactly known (due to rarity of occurrence). Few authors have suggested that color doppler imaging can be helpful in identifying the single large vitelline artery and the absence of renal arteries and 3D-sonography and MRI may complement the 2D-sonographic findings [5,6]. Fetal autopsy should be offered in all such cases so as to identify the additional major malformations and to help in understanding the pathophysiology of this condition which may offer further insight into development of human embryo as a whole.

## Conclusion

Sirenomelia is an extremely rare congenital anomaly with characteristic appearance of fused lower limbs. It has been found to have strong association with maternal diabetes. Fetal autopsy is gold standard for diagnosing additional renal, bladder and cardio-vascular malformations. Early prenatal diagnosis by first trimester scan should be the aim to minimize the trauma related to the termination of pregnancy at advanced gestation. Additionally, color Doppler, fetal MRI may further aid in the earlier diagnosis of this rare condition so as to allow first trimester termination of this fetus, rather than to face emotional trauma of second trimester abortion.

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