

Case Report

SIRENOMELIA: A CASE REPORT

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ABSTRACT

Sirenomelia or Mermaid syndrome is a rare congenital malformation characterised by fused lower limbs, genitourinary abnormalities, imperforate anus and a single umbilical artery. Its incidence is about 1:1,00,000 births. This is a case of sirenomelia which was detected in a fetus of about 35 wks. The fetus was delivered by emergency Caesarean section with poor Apgar score and died soon after birth. The fetus had fused lower limbs, a single foot and four digits. The urinary system and adrenal glands were absent. The gonads couldn't be detected. The terminal part of the colon was blind-ending and distended with meconium. The abdominal aorta below the umbilical artery was rudimentary and gave off two iliac arteries. The right iliac artery continued into the thigh. The left iliac artery was small. There was right pulmonary aplasia and left pulmonary hypoplasia. Radiograph of the fetus revealed partially fused femurs, separate tibiae, but no fibulae. There were only 3 metatarsals, each of them having a single phalanx. Sirenomelia cases usually die because of pulmonary hypoplasia, aplasia or renal agenesis. Though there are a few theories regarding aetiopathogenesis, there is still no such one which can fully explain all the features. The sirenomelia cases can be detected by ultrasound antenatally. Such babies cause difficult labor and are usually delivered by Caesarean section but death usually occurs soon after birth. Once the diagnosis is made during early pregnancy, termination of pregnancy is advisable.

KEY WORDS. Sirenomelia, Mermaid syndrome, fused lower limbs, Single umbilical artery, Malformation

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INTRODUCTION

Sirenomelia or Mermaid syndrome is a rare congenital malformation so named after the mythical Greek Siren or Mermaid. It is a condition which is characterised by fused lower limbs, genitourinary abnormalities, imperforate anus and a single umbilical artery [1]. It has an incidence of about 1:1,00,000 births [2]. Male to female ratio is 2.7:1 [3]. Sirenomelia babies are usually stillborn though cases of live births

have been reported [4]. Those which are born alive have very little chance of survival. The condition has a recurrence rate of 3-5% [5]. It is found in 2% of the mothers with diabetes mellitus [6] and monozygotic twins have 100-150 times the chance of having sirenomelia than singletons or dizygotic twins [7]. In this study, we describe a case of sirenomelia which was detected in a fetus of about 35 wks in the Department of Anatomy, Regional Institute of Medical Sciences, Imphal, India.

CASE REPORT

The fetus in the present study was picked from the specimens procured from the Department of Obstetrics and Gynaecology for PG thesis work. History revealed that a 40 year old woman with gravida 4, para 2, presented at the Department of Obstetrics and Gynaecology with cephalopelvic disproportion, severe oligohydramnios and fetal distress at 35 weeks. She delivered a 2 kg baby by emergency Caesarean section with an Apgar score of 2/10 and the baby died eventually. On gross examination, the fetus had fused lower limbs, a single foot and four digits with nails (Fig. 1). The digits on the right and left of the foot were bigger than the middle two. There was imperforate anus with a dimple at the site of the anal orifice. There was simian creases on both the palms. On internal examination of the abdomen, there were no kidneys, adrenal glands, ureters, urinary bladder, urethra or uterus. The gonads couldn't be detected. The terminal part of the colon was blindending and distended with meconium (Fig. 2). The abdominal aorta below the umbilical artery was rudimentary and gave off two iliac arteries. The right iliac artery was larger and continued into the thigh. The left iliac artery was small. It passed through an osteofibrous tunnel over the 5th lumbar vertebra but didn't reach up to the thigh. Dissection of the thorax showed right pulmonary aplasia and left pulmonary hypoplasia (Fig. 3). Radiograph of the fetus revealed partially fused femurs, separate tibiae, but no fibulae (Fig. 4). The sacrum was hypoplastic and there were hemivertebrae in the lumbar and thoracic regions. There were only 3 metatarsals, each of them having a single phalanx. The umbilical cord had only one umbilical artery (Fig. 5).

Fig. 1: Showing sirenomelia fetus before dissection.



Fig. 2: Showing colon with blind-ending distended terminal part.



Fig. 3: Showing pulmonary aplasia (right) and hypoplasia (left).

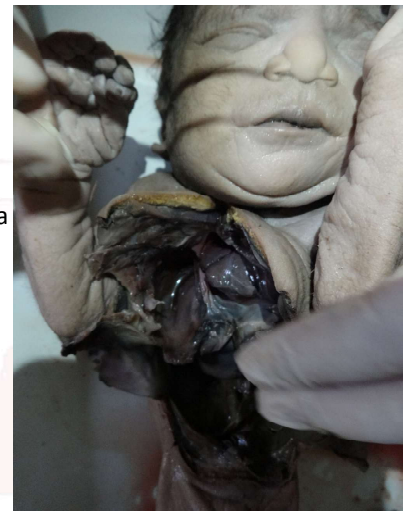
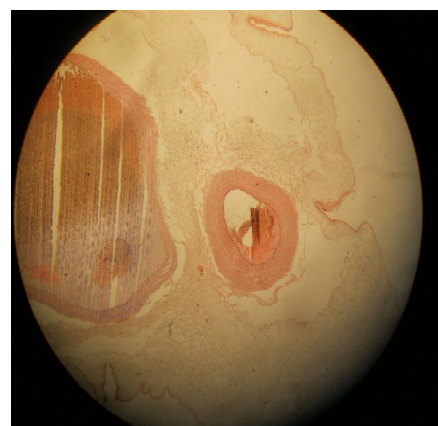


Fig. 4: X-ray showing fused femurs, separate tibiae, 3 metatarsals, single phalanges but no fibulae.



Fig. 5: Histological preparation showing single umbilical artery.



DISCUSSION

Sirenomelia babies can present with various degrees of severity. Depending on the degree of fusion of lower limbs sirenomelia can be classified into three types viz. simpus diapus (mildest type) with separate feet but fused legs; simpus unipus (moderate severity) with one foot, two tibiae, two fibulae, two femurs and simpus apus (most severe type) with no feet, one tibia, one femur [8]. According to this classification, the present case was nearest to simpus unipus type. It can also be classified into seven variants viz. Type I through VII, Type I being mildest and type VII, the most severe [9,10]. They are type I with separate thigh and leg bones and soft tissue fusion; type II with single/fused fibula but femurs but tibia are separate; type III with absent fibula but femurs and tibiae are separate; type IV with partially fused femurs, fused fibulae and femur but tibiae are separate; type V which is same as type IV but without fibulae; type VI with single femur and tibia but with no fibula; type VII with single femur without tibia or fibula. The fetus in the present case belonged to Type V of Stocker and Heifetz [9].

The most striking feature of sirenomelia is the various degrees of fusion of the lower limbs. The clinical conditions with overlapping phenotypic features are caudal regression syndrome and VACTERL syndrome. Caudal regression syndrome is characterised by developmental abnormalities of the lower limbs and sacrum associated with spinal cord abnormalities and urinary incontinence. In caudal regression syndrome, two umbilical arteries instead of one are found and fused lower limbs, renal agenesis and imperforate anus are rare [11,12]. VACTERL syndrome is characterised by vertebral anomalies, anal atresia, cardiac defects, tracheo-oesophageal fistula, renal and radial anomalies and limb defects. It shares many features with sirenomelia but the defects are more severe in the case of sirenomelia than in VACTERL syndrome [13]. The etiopathogenesis of sirenomelia is not clearly known. Two important theories which have been proposed are the defective blastogenesis [14] and vascular steal [15]. The defective blastogenesis theory states that sirenomelia results from the defective

gastrulation in the early stage of embryonic development which leads to defective migration and differentiation of the caudal somites. The vascular steal theory describes that diversion of blood supply by a single vitelline artery from the caudal region to the placenta causes a nutritional deficit in that area. The single umbilical artery found in sirenomelia is the persistent vitelline artery that assumes the function of the absent umbilical arteries [10]. The abdominal aorta below origin of the single umbilical artery is usually absent [10]. In the present case, the lower part of the abdominal aorta was rather hypoplastic and only the right iliac branch continued into the thigh. The argument against the vascular steal is that it cannot explain the abnormalities in the distant areas such as the lung, cardiac, cranial or esophageal defects in sirenomelia cases [16]. In addition to the defects in the bones of the lower limbs, there may be abnormal development of pelvis or vertebrae [14]. In the present case, hemivertebra was found in the thoracic and lumbar regions and the sacrum was hypoplastic. In sirenomelia, the kidneys and urinary tracts are usually absent [14]. In the present case, the whole urinary system was absent. There was pulmonary aplasia on the right side and pulmonary hypoplasia on the left side. Sirenomelia cases usually die because of pulmonary hypoplasia or aplasia or renal agenesis [10]. In pregnant women, diagnosis is usually done with ultrasound. But it is not easy to diagnose this condition and they can be missed with this procedure because of the associated oligo- or anhydramnios [10].

The sirenomelia cases may be detected antenatally by ultrasound or detected for the first time at the time of labour in those women who didn't have antenatal check-ups. Sirenomelia babies cause difficult labor and are usually delivered by caesarean section but death usually occurs soon after birth.

CONCLUSION

Many cases of sirenomelia have so far been reported but there is lack of evidence about the etiopathogenetic mechanism especially the one which can explain all the features of sirenomelia. Sirenomelia cases are characterised

by poor prognosis because of pulmonary or renal aplasia. Early antenatal diagnosis, especially during the first trimester, is advisable. Once the diagnosis is made, termination of pregnancy is the best thing that can be done as treatment of this condition is unsatisfactory.

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Conflicts of Interests: None

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