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**Case Report**

# Sirenomelia, the Mermaid syndrome: a case report

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## ABSTRACT

Sirenomelia, also known as mermaid syndrome, is a rare congenital anomaly characterized by a single lower extremity which is associated with abnormalities in other organ systems, commonly affecting the gastrointestinal and the urogenital systems. It is sporadic with no increased risk in subsequent pregnancies. In almost all the cases of sirenomelia, a single umbilical artery (SUA) is present which arises from the abdominal aorta. The exact etiology of sirenomelia is unknown. Sirenomelia can be confidently diagnosed in the 1st trimester while the diagnosis in the 2nd and 3rd trimesters is difficult due to the lack of amniotic fluid in the later gestation. Antenatal diagnosis of this universally lethal condition is desirable so that possible termination of pregnancy can be offered at the earliest.

**Keywords:** Mermaid syndrome, Oligohydramnios, Sirenomelia, Sirenomelia sequence

## INTRODUCTION

Sirenomelia is a birth defect of the lower body characterized by the apparent fusion of the legs into a single lower limb.[1] Sirenomelia sequence has a characteristic external phenotype along with multisystem involvement of predominantly uro-genital, pelvic and gastrointestinal malformations.[1] Sirenomelia is usually incompatible with life due to the serious visceral abnormalities with high mortality in the perinatal period.[1] Stillbirth occurs in more than 50% of cases of sirenomelia.[2] This condition is 100 times more likely to occur in identical twins than in single births or fraternal twins.[2] Complete or partial bilateral renal agenesis is common and most often incompatible with life, although there have been very rare reported cases of patient survival secondary to the presence of a normal kidney.[3] Occasionally upper body visceral malformations have also been reported in various case reports.[1] Here, we describe one such rare case of sirenomelia born in our hospital.

## CASE REPORT

A 26 year old G3P2L2 mother, registered at 7 months of gestation, without any significant past or antenatal history, delivered a 1505 gram malformed baby at 32weeks of gestation through lower segment caesarean section without any intrapartum or postpartum complications. Baby was resuscitated and shifted to NICU for poor Apgar scores with cyanosis, global hypotonia, heart rate <60 beats/minute and irregular respiration at birth. Antenatal examination before delivery revealed a fundal height corresponding to 28 weeks of gestation with palpable fetal parts on abdominal examination, suggestive of oligohydramnios and intrauterine growth retardation (IUGR). Antenatally, her routine investigations including blood sugar, ultrasonography reports and congenital anomaly scan done were normal. There was no history of any antenatal teratogenic drug intake. She had not received any iron or

folic acid supplements during pregnancy. There was no family history of consanguinity or any congenital anomaly. On examination baby had characteristic features of sirenomelia sequence with completely fused lower limbs into a single antero-posteriorly flattened limb, with dorsum of the single foot facing posteriorly and four toes, upper limbs being normal. Baby had Potter facies including flattening of the nose, receding, soft flat low set ears with apparent absent cartilage and hypertelorism. Additional malformations like deformed pelvic bones, single umbilical artery (SUA), sacral dimple, genital tag, absent external genitalia as well as urethral and anal opening were noticed in the baby (Figure 1 and 2).Our baby died 30 minutes after birth from respiratory distress most probably due to pulmonary hypoplasia and renal agenesis. Post-natal x-rays revealed hypoplastic lungs, bell-shaped chest, paucity of distal abdominal gas shadows, hypoplastic pelvic bones, partially fused femur bones, two tibiae, absent fibulae, metacarpals and phalanges of a foot seen (Figure 3) Autopsy was denied by the family. The family was counselled to follow up for antenatal genetic counselling during the next pregnancy.

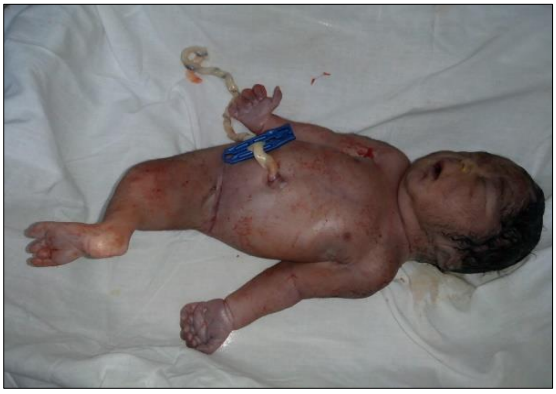

**Figure 1: Clinical features of baby with Potter's (Anterior view) showing potter facies.**

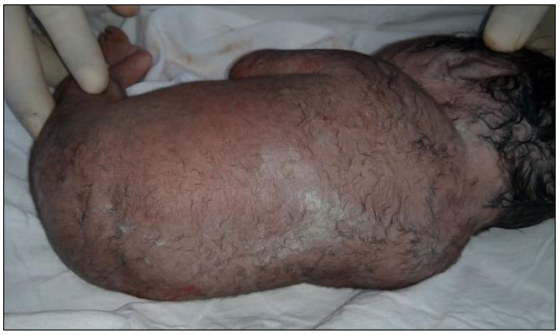

**Figure 2: Clinical features of baby with Sirenomelia (Posterior view) showing absent anal opening.**

Figure 1 shows Clinical photograph of baby with sirenomelia (Anterior view) showing Potter facies: flattening of the nose, receding, soft flat low set ears with apparent absent cartilage, hypertelorism. Lower limbs are completely fused into a single antero-posteriorly flattened limb, posteriorly facing dorsum of single foot, four toes, normal upper limbs, genital tag, absent urethral opening.

Figure 2 shows clinical photograph of baby with sirenomelia (Posterior view) showing flexion of body at hip joint, sacral dimple, absent anal opening.

Figure 3 represents roentogram of baby with Sirenomelia shows hypoplastic lungs, bell-shaped chest, paucity of distal abdominal gas shadows, hypoplastic pelvic bones, partially fused femur bones, two tibiae, absent fibulae, metacarpals and phalanges of a foot.

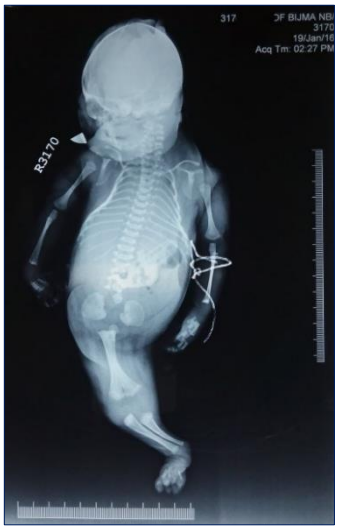


**Figure 3: Roentogram of baby with Sirenomelia.**

## DISCUSSION

Sirenomelia is a limb anomaly in which the normally paired lower limbs are replaced by a single midline limb reported in all ethnic groups around the world.[1,4] The incidence of human sirenomelia varies between 1.1 and 4.2 per 100,000 births.[1] The sex in sirenomelia was generally based on gonadal or, rarely, on chromosomal examination, which explains why almost half of the cases had undetermined sex.[3,4] Sirenomelia is a sporadic condition, with normal fetal karyotyping without an increased risk of recurrence in subsequent pregnancies.[5] Genetic as well as environmental factors (nutritional and toxic) like retinoic acid, ochratoxin A, cadmium, lead and cocaine have been reported as causes for sirenomelia.[1,4]

Sirenomelia was named after the creatures with the head of a woman and body of a bird from the wings downwards called ‘sirens’ described in Greek mythology, who narcotized the sailors with their enchanting music and voice to later kill them.[1] Ironically, the sirenomelia human malformation is a severe condition despite the present perception of sirens being portrayed overtime as romantic beautiful aquatic creatures with mermaid-like appearance.[1] Sirenomelia is also described as ‘mermaid syndrome’ which includes fusion of the lower limbs,

right renal agenesis, deformity of the bony pelvis, single umbilical artery, absent external genitalia, and imperforate anus.[2]

Based on the presence or absence of feet, sirenomelia was previously classified, as sympus apus or sirenomelia (no distal foot element), sympus monopus or uromelia (1 foot) and sympus dipus or symmelia (2 feet) which is now obsolete.[1] Potter's syndrome consisting of Potter's facies, oligohydramnios and pulmonary hypoplasia, as was seen in our baby, is almost invariably presents with bilateral renal agenesis.[2] The recent Stocker and Heifetz classification of sirenomelia (Table 1) based on the presence of skeletal elements in the thigh and leg ranges from type I, the mildest form, with all bones present in the two fused limbs, fusion affecting only superficial tissues to type VII, the most severe form, with only 1 single bone present, and no indication of legs or feet.[1] Our baby with presence of one foot fitted into Type V characterised by partially fused femur bones with two unfused tibiae and absent fibulae corresponding to sympus monopus or uromelia of the previous classification.

The pathophysiology of sirenomelia includes approximation and merging of the hind limbs beginning from the posterior side, which is possible if the parts lying between them, like midline structures like cloaca and urogenital sinus on ventral side and somites and neural tube on the dorsal side, fail to develop resulting in the dysmorphology.[1] The normal limb development, a continuous process involving the mesodermal and ectodermal components, is divided into four stages: 1) the bud (outgrowth), 2) the paddle (dorso-ventral flattening), 3) the plate (relative expansion of the distal end), and 4) the rotation (around the proximo-distal axis). This results in the ventral surface eventually facing dorsally as the hind limb buds rotate around their proximo-distal axis medially, which is halted due to early abnormal fusion of the limbs along their posterior margins resulting in soles facing anteriorly, abnormal flexion of the knees and fibulae adopting a medial position between the tibiae.[1,4]

**Table 1: Classification of Sirenomelia by Stocker and Heifetz (1987).**

| Type | Characteristics |
|---|---|
| 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 |

Various visceral malformations commonly seen in sirenomelia are: 1) renal including total renal agenesis, ectopic renal tissues, 2) absent external genitalia or genital tag of tissue; gonads remaining unaffected and 3) gastrointestinal like blind ending colon, renal atresia, imperforate anus.[1,2] The other associated malformations are lumbosacral and pelvic like sacral agenesis, malformed vertebrae and hemi-vertebrae. Occasionally upper body malformations, of uncertain mechanism, like cleft palate, upper thoracic and cervical vertebral anomalies, CNS anomalies, pulmonary hypoplasia and cardiac defects are also seen associated with sirenomelia.[1]

The vascular malformations seen in sirenomelia are due to the disturbance in the development of the vitelline and umbilical arteries. The commonest abnormality seen is single umbilical artery (SUA) of abnormal origin branching off high after the celiac branch from abdominal aorta which then abnormally narrows down and lacks a considerable number of branches resulting in impaired perfusion to the lower extremities, kidneys, urogenital tract and large intestine.[1,5,6] The pathognomonic feature of sirenomelia is this SUA of abnormal origin referred to as the persistent vitelline artery (PVA), a possible derivation from the vitelline plexus thus distinguishing it from other cases of SUA, found in about 1% of newborns, having a normal origin not related to other malformations.[1,6]

The 2 main pathogenic hypotheses based on various clinical studies explaining the possible causal mechanism of sirenomelia are the vascular steal phenomena and the blastogenetic hypothesis.[1,2] The other mechanical hypotheses are lateral compression of the caudal body by amniotic folds and medial compression by over-distension of the neural tube.[1]

The vascular steal hypothesis states that SUA of vitelline origin diverts blood flow to the placenta from the aorta thus causing a severely deficient circulation of the caudal mesoderm and the variable development of sciatic artery, a branch of recurved distal part of dorsal aorta initially supplying the leg bud. This leads to the agenesis of lower midline structures which along with the subsequent abnormal approximation of both lower limb fields is responsible for the variable phenotype seen in sirenomelia.[1,2,5,6]

According to the defective blastogenesis hypothesis, sirenomelia is believed to be secondary to the maldevelopment of the caudal somites and tail bud, due to primary defect in blastogenesis, occurring in the final stages of gastrulation (at about the 3rd week of gestation) in humans, concomitantly affecting the organ and vessel development.[1,5] The phenotypic variability depends on the intensity, time of initiation and duration of the underlying event.[1,5] A cascade of events occurs during gastrulation in vertebrates as the initial 2-layered blastocyst develops into a 3-layered embryo with formation of end mesoderm in a rostro caudal sequence. At late gastrulation stage, the remnant of the regressing primitive peak forms the caudal eminence (tail bud). This forms a thick ectodermal area known as the ventral ectodermal ridge (VER) which undergoes an epithelial-

mesenchymal transition process, accounting for the accumulation of mesoderm in the lateral and ventral tail bud region.[1,2,5]

Caudal regression syndrome (CRS) or caudal dysgenesis (CD) are broad terms that refer to a heterogenous constellation of congenital caudal anomalies affecting the caudal spine and spinal cord, the hindgut, the urogenital system, and the lower limbs.[7] The constant presence of an abnormal SUA of vitelline origin, occurrence of cases without dorsal defects of the neural tube and spine, along with a lack of a clear association with maternal IDDM differentiates sirenomelia from CD.[1,7,8] In addition, in CD, the lower limbs are not fused and liquor is usually adequate.[5] The categorization of VACTERL syndrome required the presence of at-least 3 of these defects: vertebral, anal, cardiovascular, tracheo-oesophageal, renal and limb dysgenesis.[6,9-11] Inspite of having 3 anomalies qualifying for VACTERL, the diagnosis in our baby goes more in favour of sirenomelia due to the single fused limb being the striking feature. The difference between sirenomelia sequence and VACTERL lies mainly in the severity of the component defects, with the single lower limb in sirenomelia being an epitome of other severe malformations.[6,9] Clinical data suggests that, sirenomelia, like other malformations affecting multiple organs, arises very early during development due to an event or disturbances of a basic single developmental mechanism which could explain the abnormal vascular pattern as well as the variable organ hypoplasia.[1]

A genetic basis, can be explained based on studies like, 1) disruption of Cyp26a1 enzyme causing a gain of retinoic acid (RA) function, 2) reduction of Bmp signalling in the ventral caudal embryonic mesoderm leading to occurrence of several caudal defects, severe cardiac malformations and absence of major vessels. RA decreases bone morphogenetic protein (Bmp) signal duration and reciprocally, Bmp signalling negatively regulates RA signalling during chondrogenesis, RA and Bmp thus regulating each other.[1,2,4]

Sirenomelia is diagnosed on antenatal (ANC) ultrasonography (USG) at 18 week gestation by 1) oligohydramnios, 2) bilateral renal agenesis 3) fusion of the lower extremities with limited value in the 2nd and 3rd trimesters due to renal agenesis causing severe oligohydramnios; however SUA, fused femur, decreased distance between two femurs, decreased/absent motility of the lower extremities, absent urinary bladder, undetermined genitalia, lumbosacral agenesis, and anorectal atresia may clinch the diagnosis.[3,5,12] USG is useful to offer the option of termination of pregnancy to the parents prior to 20 weeks of gestation.[2,5,12] Features of sirenomelia are easily demonstrated on prenatal MRI even in the presence of oligohydramnios.[3,5]

Sirenomelia is fatal in most cases secondary to the pulmonary hypoplasia caused by the severe oligohydramnios due to renal agenesis. Very few cases survive beyond the neonatal period due to the presence of a functioning renal system without total obstructing renal failure. Extensive surgeries are needed for anorectal and genitourinary abnormalities and limb reconstruction in the babies who survive.[5]

## CONCLUSION

Sirenomelia is a lethal sporadic congenital anomaly with a poor outcome and an unknown exact etiology. Transvaginal USG and colour doppler have a key role in making the diagnosis with 3D ultrasound helping in better characterization of the abnormality. There is scope for further studies to be done on the molecular level based on the postulated clinical hypotheses to determine the exact causal factors of sirenomelia.

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