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Case report

# Sirenomelia associated with discoid adrenal and lumbar meningocoele: An autopsy report

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ABSTRACT

Mermaid syndrome or Sirenomelia is a rare congenital deformity in which the legs are fused and bears resemblance to mermaid's tail. It carries a poor prognosis, due to associated urogenital and gastrointestinal abnormalities. An early antenatal diagnosis using Magnetic Resonance Imaging (MRI) can help in termination of pregnancy. Embryologically, it is considered as the extreme form of caudal regression syndrome due to the persistence of vitelline artery. Here, we report a case of Sirenomelia associated with bilateral renal agenesis along with the rare findings of discoid adrenal, lumbar meningocoele and abnormalities of the hand.



## 1. Introduction

Sirenomelia or "mermaid sequence" is an extremely rare and lethal sequence of congenital developmental disorders characterised by anomalies of the lower limbs and the lower spine [1]. It is a severe form of caudal defect with an incidence of 1 per 100,000 births with a male to female ratio of 3:1 [2]. It was first described by Rocheus in 1542 and by Palfyn in 1553 [1]. According to Stevenson, sirenomelia is a limb anomaly in which the normally paired lower limbs are replaced by a single midline limb. Affected infants are born with partial or complete fusion of the legs. Vascular steal phenomenon remains the most important theory in its pathogenesis [2,3]. The striking external phenotype associated with a variable combination of severe visceral anomalies makes Sirenomelia a multisystem developmental disorder. The commonly associated malformations are agenesis or dysgenesis of the kidneys, absent external genitalia, imperforate anus, lumbosacral and pelvic bone abnormalities and a single umbilical artery (SUA) [2,4]. Central Nervous System (CNS) anomalies have been reported in less than 10% of sirenomelic infants [5].

Here we present a case of sirenomelia with absence of hand digits in one of the upper limbs and a fused single lower limb. Autopsy revealed several abnormalities in internal organs including the rare findings of discoid adrenal and lumbar meningocoele.

## 2. Case report

A 37 weeks old preterm infant of unidentified gender was born to a 19 years old primigravida mother with no significant past medical history. She was not diabetic or hypertensive. Antenatal ultrasonographic scan (USG) revealed a single fetus with features of intrauterine growth retardation and severe oligohydramnios. The infant was delivered by emergency caesarean section, indication being oligohydramnios with small baby. The baby did not cry at birth. The Apgar score was 6 and 7 at 1 and 5 min, respectively. The baby died after 5 h. On physical examination of the newborn, the crown rump length was 20 cm and its weight was 1250 g. The face showed dysmorphic facies with mild hypertelorism, low set big ears, receding chin and short neck like Potter's facies. External examination revealed an imperforate anus and absence of external genitalia. A skin tag was noted in the lumbar region (Fig. 1b and c).

At autopsy, the upper limbs were well developed but the right hand was flipper like with no bones. There was a single lower limb with flipper like foot where only the great toe was identified. Postmortem radiograph revealed the presence of one femur and one tibia (Fig. 1a). The fibula was absent. Externally there was kyphoscoliosis. There was absence of both the kidneys, ureters and urinary bladder. No gonads were found. The gastrointestinal system ended in a blind loop (Fig. 1d). The lungs were hypoplastic and weighed 8 g (Fig. 3d). The umbilical cord was noted to have a single umbili-

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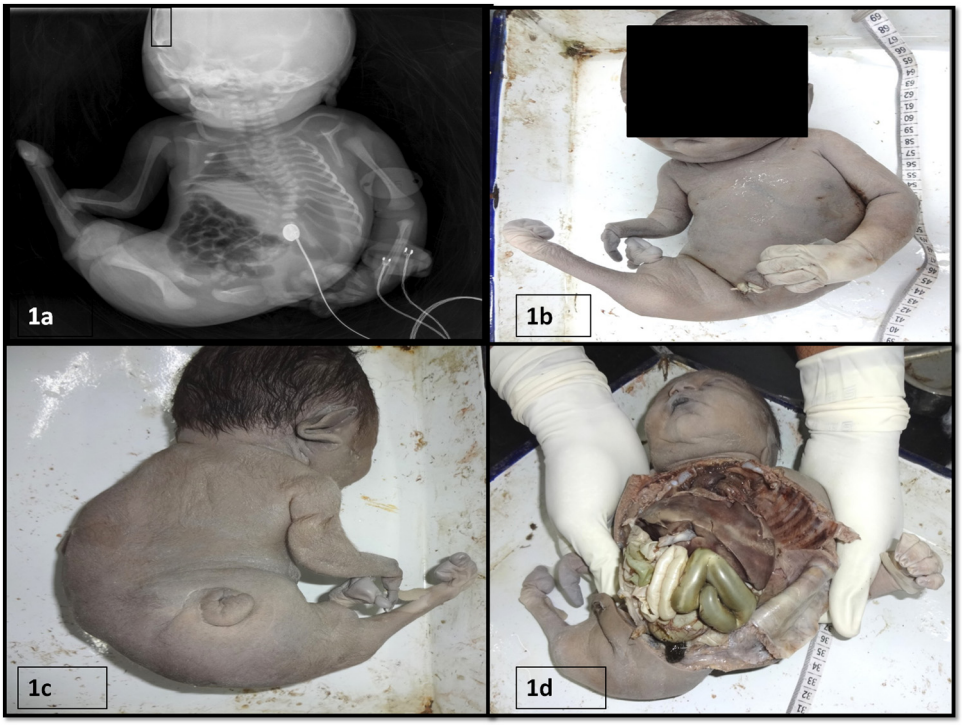


**Fig. 1.** (a) X-ray (Antero Posterior view) of the new born showing one femur and one tibia. (b) Gross photomicrograph of the new born at autopsy showing single lower limb and no external genitalia. (c) Photomicrograph showing skin tag and absence of anus. (d) Blind ending gut, in situ organs.

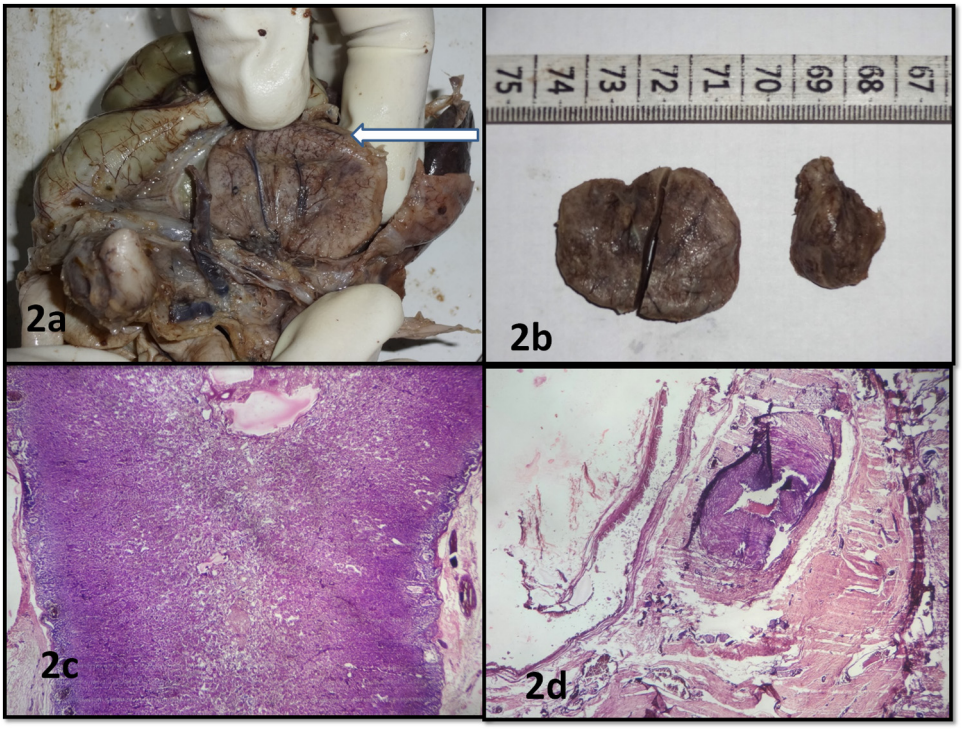


**Fig. 2.** (a) Photomicrograph showing discoid adrenal in situ, marked with arrow. (b) Discoid adrenal right sided and normal appearing adrenal. (c) Photomicrograph showing low power view of discoid adrenal (H & E stain). (d) Photomicrograph showing single umbilical artery.

cal artery (Fig. 2d). The inferior mesenteric artery and renal arteries were absent. The abdominal aorta gave rise to the single umbilical artery. The right sided adrenal gland was like a flat disc and the left sided adrenal was semilunar in shape (Fig. 2a–c). On microscopic examination,the adrenals were unremarkable. Microscopic examination of the lumbar skin tag revealed presence of neural tissue capped by leash of small hemangiomatous vessels indicating