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Case Report

Sirenomelia: Mermaid Syndrome – A Rare Autopsy Case Report

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Abstract

Sirenomelia, the Mermaid Syndrome is a rare and lethal congenital anomaly with an incidence of one in 60,000 pregnancies. Sirenomelia is characterized by fusion of the lower limbs, commonly associated with renal agenesis, absent external genitalia and other gastrointestinal defects. We report a case of sirenomelia in a stillborn 26 wks fetus received for autopsy. Apart from the characteristic features it was also associated with single umbilical artery, potter's facies and hypoplasia of various internal organs. Ultrasound may be useful in the early antenatal detection of this anomaly however coexisting oligohydramnios as in this case makes the visualisation of caudal extremity difficult. Early prenatal diagnosis should be the aim to minimize the trauma related to the termination of pregnancy at advanced gestation.

Keywords: Sirenomelia; Mermaid syndrome.

Introduction

Sirenomelia is a rare birth defect also known as 'Mermaid syndrome' due to characteristic fusion of both lower extremities which look like Mermaids tail, found only in one in 60,000 live births.[1] Up till now, only 300 live births have been reported and only two of them are alive at present.[2] It is more frequent in males and in one of identical twins.[1] Sirenomelia is usually associated with severe anomalies like bilateral renal agenesis which is incompatible with life in majority of cases. The presence of oligohydramnios due to bilateral renal agenesis usually hinders adequate ultrasonographic exploration of caudal extremity of the fetus hence diagnosis is usually made at autopsy.[3] Here we present a rare case of sirenomelia with multiple associated anomalies diagnosed on autopsy.

Case History

A 24 yrs old primigravida presented with complaints of abdominal pain and history of 5mths amenorrhoea. There was no prior antenatal checkup or ultrasonography. There was no significant medical history. Emergency ultrasound revealed oligohydramnios and bilateral renal agenesis. She delivered a dead fetus of 26 wks of gestational age weighing 700 gms, which was received for autopsy.

Post-mortem X-ray revealed sacral agenesis,

Figure 1: X-Ray Showing Single Femur and Redundant Tibia

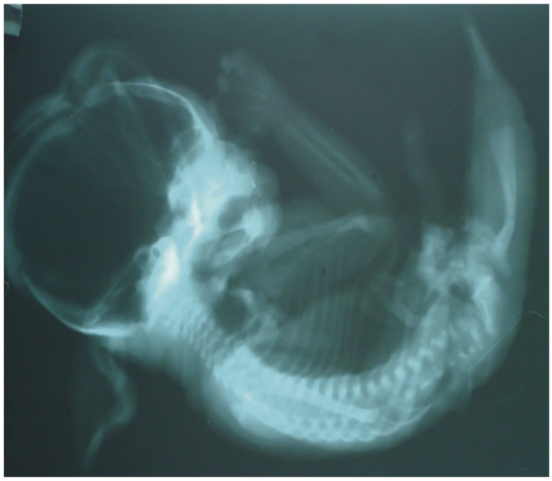

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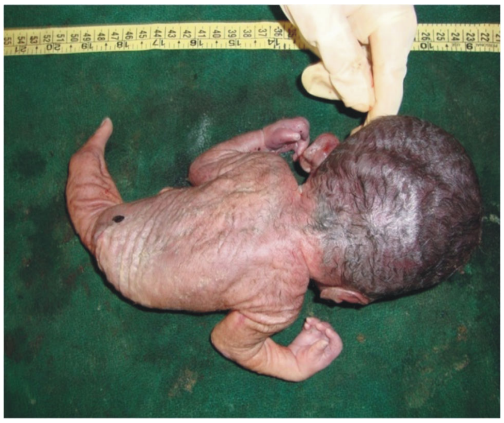
Figure 2: External Examination Revealed Fusion of Both Lower Limbs Forming One Segment without Feet (Sirenomelia Apus)



single femur and redundant tibia (Figure 1). External examination revealed fusion of both lower limbs forming one segment without feet, looking like a tail of 12 cms length. The above findings were consistent with the diagnosis of Sirenomelia apus (Figure 2). Single umbilical artery (Figure 3), potter's facies, cutis laxa, imperforate anus, external genitalia agenesis were also noted.

Figure 3: Single Umbilical Artery

Figure 4: Blind Pouch of Rectum

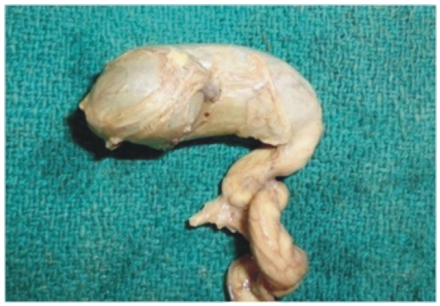
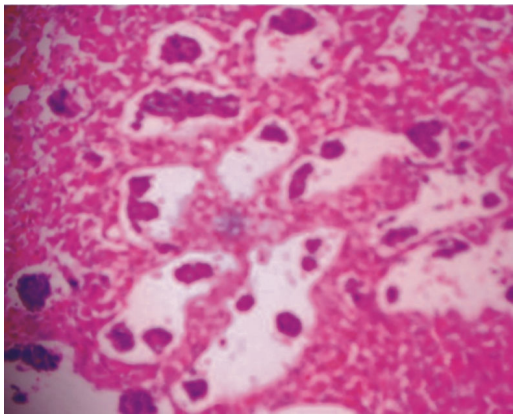


Figure 5: Microscopy Showing Abortive Tubules in Adrenals (H&E 400x)



In situ examination revealed non-aerated hypoplastic lungs, intestinal diverticulum, blind pouch of rectum (Figure 4), bilateral renal agenesis and urinary system agenesis. Testis like structures found in lower abdomen (confirmed microscopically). Microscopically adrenal tissue revealed few abortive tubules (Figure 5).

Discussion

Sirenomelia Sequence or Mermaid Syndrome was originally described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens4. It is a severe developmental field defect of the posterior axis caudal blastema, resulting in apparent fusion of the lower limb buds1. Around 300 cases reported in the world literature, of which 13 have been from India.[2]

Sirenomelia is classified depending on extent of fusion of lower extremities as 1) sirenomelia apus: no feet, only one tibia and one femur, 2) sirenomelia unipus: one foot, two femora, two tibiae, two fibulae and 3) sirenomelia dipus: two feet and two fused legs.[5] This case was of sirenomelia apus. Stocker and Heifetz classified sirenomelia into type I to type VII, according mainly to the presence of skeletal elements in the thigh and leg. In type I, the mildest form, all bones in the two fused limbs are present, and the fusion only affects superficial tissues. In type VII, the

most severe form, only a single bone is present, with no indication of legs or feet.[6]

Various theories have been postulated to explain the etiology of sirenomelia. Theories range from intrauterine force, failure of development of caudal somites, injury to the caudal mesoderm between 28-32 days of fetal development, neural tube overdistension in the caudal area to the more recent vascular steal theory.[7] Vascular stealing due to presence of single large umbilical artery leading to abnormal ischaemic development of caudal end of the embryo is one of the most favoured theory. Single umbilical artery was also noted in this case.

Other anomalies in the present case included potter's facies (large, low-set ears, prominent epicanthal folds, hypertelorism, flat nose, and receding chin)8, cutis laxa, imperforate anus, external genitalia agenesis, non-aerated hypoplastic lungs, intestinal diverticulum, blind pouch of rectum, bilateral renal agenesis and urinary system agenesis.

Sirenomelia was earlier thought to be a form of caudal regression syndrome, however it is reclassified to be considered a separate condition. There is a strong association of Sirenomelia and Caudal Regression Syndrome with maternal Diabetes. Although in this case the mother did not have evidence of this risk factor but should be counselled about early screening in the subsequent pregnancy. Several anomalies are common to both conditions, but presence of an aberrant abdominal umbilical artery/"persistent vitelline artery" has been invoked as the chief anatomic finding that distinguishes Sirenomelia from Caudal Regression Syndrome.[4]

The sirenomelia is diagnosed by sonography as early as 9 weeks. Diagnosis is difficult during the second trimester because of the severe oligohydramnios.[2] In our case, the diagnosis was primarily of bilateral renal agenesis and oligohydramnios during scan. The lower limbs were not seen properly due to severe oligohydramnios.

Survival depends on the associated anomalies, especially renal function. Treatment includes supportive care and multidisciplinary surgical approach. Owing to visceral abnormalities, sirenomelia is usually incompatible with life; death occurs in the perinatal period.[2] At present only two have survived for years since birth with the oldest one being 25 yrs old now.

Conclusion

Sirenomelia is a rare multisystem congenital malformation of unknown etiology. This is a rare autopsy case of sirenomelia apus with associated agenesis and hypoplasia of diverse internal organs. Early diagnosis of this fatal anomaly is useful so that the option of pregnancy termination may be given to the parents and to minimize the trauma related to the termination at advanced gestational age.

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