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Case Report

Paediatrics Section

Message from the Mermaid-Do not Miss the Anomaly Scan

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ABSTRACT

Sirenomelia, also known as Mermaid syndrome is a rare congenital structural anomaly, caused by a vascular insult during the embryonic life. It is characterized by varying degrees of fusion of the lower extremities along with urogenital and gastrointestinal malformations. Diagnosis of sirenomelia can be made as early as 11-13 weeks and hence the birth of this syndromic baby can be prevented. We report a case of Sirenomelia- Mermaid Syndrome along with the clinical autopsy findings and review its literature.

Keywords: Breech, Oligohydramnios, Sirenomelia, Urogenital malformations

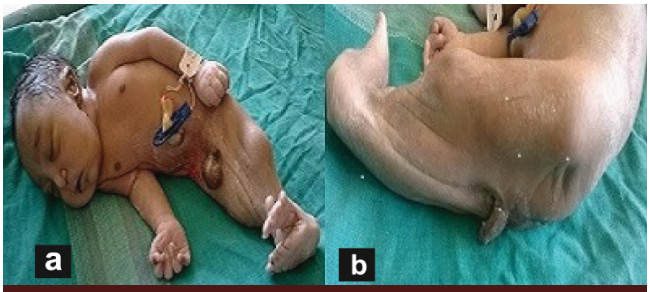
CASE REPORT

A 25-year-old primigravida presented at 39 weeks period of gestation in early labour with breech presentation. She was an unbooked case. Her antenatal period was uneventful. All investigations were within normal limits. A third trimester ultrasonography, which was the only single sonography report with her, suggested severe oligohydramnios and breech presentation. Due to decreased amniotic fluid and advanced gestational age, congenital anomalies could not be excluded. She was non diabetic, non alcoholic and non smoker. There was no history of any congenital anomalies in the family, any radiation exposure or known exposure to any teratogen. It was a non consanguineous marriage.

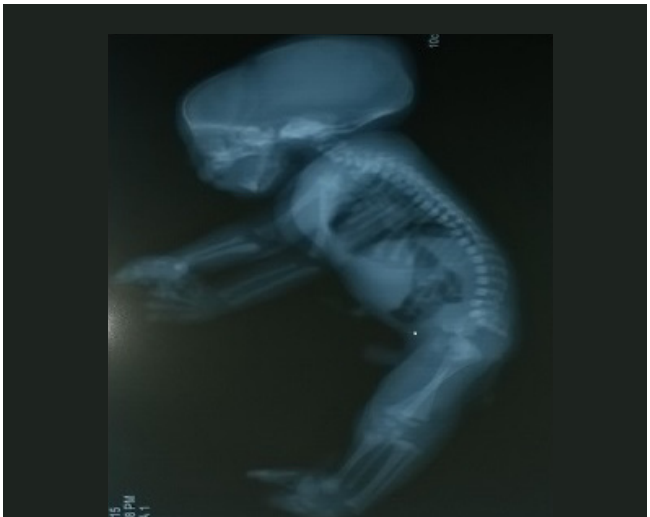
An emergency caesarean section was done in view of foetal distress but resulted in the birth of an anomalous baby with APGAR score of 3/10 at 1 minute. The baby expired within half an hour, due to respiratory distress, despite resuscitation.

On examination of baby, it had Potter's face, fused lower limbs up to the ankle, two feet with one toe missing in the right foot, empty scrotal sac and no penis, no anal opening and a tail like projection on the dorsal surface at caudal end [Table/Fig-1a,b]. The weight of the baby was 2.25 kg. The placenta was normal and the umbilical cord had a single umbilical artery.

A postmortem infantogram showed the presence of all the three lower limb bones. Upper limb bones were normal. There were no vertebral anomalies [Table/Fig-2]. On autopsy, the lungs were hypoplastic and the liver was enlarged. There was renal, vesicle and ano-rectal agenesis with meconium filled blind sigmoid colon [Table/Fig-3a]. Heart, diaphragm, stomach spleen and other organs were normal in position and morphology. The



[Table/Fig-1]: (a) Potter's face and fused lower limbs; (b) Dorsal view showing anal imperforation & a tail like projection.



[Table/Fig-2]: An infantogram showing all the bones in lower limb.

histopathological examination of sections of umbilical cord revealed single umbilical artery [Table/Fig-3b]. All the features were consistent with the diagnosis of sirenomelia.

[Table/Fig-3]: (a) Autopsy showing absent kidneys and meconium filled sigmoid colon; (b) Single umbilical artery on histopathology.

DISCUSSION

A mermaid is a mythical marine creature with the head and upper body of a woman and the lower body resembling the tail of a fish. Mermaids are assigned the biological order sirenia comprising marine animals like dugongs and manatees. Hence, this syndrome is named Sirenomelia-Mermaid Syndrome [1]. It is a rare congenital malformation, not compatible with life, with an incidence of 1 in 1, 00,000 live births. It is more common in twins especially monozygotic and has a male preponderance (M: F= 3:1) [2]. However, in our case it was a singleton pregnancy. It is characterised by fused legs and is usually associated with urogenital and gastro-intestinal malformations. The defect varies from simple cutaneous fusion of the limbs to the absence of all long bones but one femur. In mild variety, the baby has two limbs fused into one, only to the extent of the skin. The feet may be fully formed and in many cases, are only attached at the ankles (Sympus Dipus). All the three main bones of the leg are fully and correctly formed. In this situation, a small surgery can easily correct the deformity. The severe variety is very difficult to manage. There may be a single leg which may be complete or incomplete (Symelia/Sympus Apus). There is complete absence of foot structures and out of the three long bones, only two are present in the entire limb [3,4]. In Symelia Unipus, all the ten toes are derived from one foot.

There may be shortened and deformed spine with abnormal and absent vertebrate. A meningo-myelocoel may be present. The pelvic bones may be partially absent. There are either dysplastic kidneys with small cysts or bilateral renal agenesis, absent bladder and absent or ambiguous genitalia. Oesophageal atresia, abdominal wall defects, cardiac anomalies, imperforate anus and severe oligohydramnios or anhydramnios may also be present. The condition is almost always fatal. Fifty percent of the children are born alive, but mostly succumb to death in the first neonatal week [5].

Various theories for this anomaly have been described but none has been able to explain its definite cause. This syndrome still remains a mystery. Embryologically, it is hypothesised that there is some insult of the caudal mesoderm between days 28 -32 of fetal life which results in the sirenomelia sequence [6]. The primitive streak at the lumbar and sacral areas undergoes a developmental arrest, therefore the midline structures i.e., cloacal and midline derivatives are destroyed. 'What causes this embryological insult' is still not clear.

Another theory regarding vascular steal phenomenon seems more appealing. According to this, there is a single large umbilical artery that enters aorta in the upper abdomen. Therefore there is vascular compromise in the caudal region resulting in redistribution of blood flow and nutrients from all the caudal structures of the embryo towards the placenta. This depletion of nutrients causes further developmental arrest or malformation of the caudal structures [7]. In the present case also, presence of single umbilical artery supports the Stevenson's theory [7].

Although, more than 300 cases of sirenomelia have been reported so far, the purpose of reporting this case is to emphasize the importance of anomaly scan so that early diagnosis can be made and such foetuses can be terminated timely. There are reported cases in which the diagnosis of sirenomelia has been made as early as 11-13 weeks and upto 20 weeks [8,9]. It is usually difficult to make a definite diagnosis of Sirenomelia in the third trimester owing to severe oligohydramnios due to bilateral renal agenesis as it happened in our case. However, during the early second trimester the amount of amniotic fluid may be sufficient enough to allow diagnosis [10]. Doppler flow imaging can detect a two vessel cord, which is a frequent association. A radiologist should therefore actively look for the sonographic features of sirenomelia (oligohydramnios, single lower limb, absent bladder, undetermined external genitalia, anorectal atresia, lumbosacral agenesis), in the early anomaly scan and early second trimester scans of all fetuses with bilateral renal agenesis, malformed lower limbs and a single umbilical artery, which can allow pregnancy termination at an early stage [11].

In developing nations like ours, still a large number of women miss the crucial anomaly scan due to lack of awareness and knowledge. They just present with a single third trimester scan which hardly benefits in such cases. Had there been a prenatal diagnosis in this case, we could have avoided the caesarean section.

CONCLUSION

Congenital anomalies are still considered a curse at many places and are sought to be cured by in human practices like witch craft which have no place in the modern times. As diagnostic modalities get refined, our capabilities of identifying the birth defects improve. But at the same time it is important that tools like print and electronic media are utilised for the generation of

awareness in the general population so as to prevent the birth of an anomalous baby and subsequent psychological outcomes. Thus, we must consider it a message from the mermaid that no pregnancy should miss an anomaly scan.

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