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Sirenomelia: Mermaid Syndrome: A Case Report

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Abstract

Sirenomelia (mermaid syndrome) is a very rare congenital anomaly which results in fusion of two lower limbs along with manyother visceral malformations. We report a case of mermaid syndrome in Maharashtra, India. An 18 – 20 weeks old abortus with lower limbs fused together like mermaid appearance. The abortus had fused lower extremities but bony structures for each leg were present on x- ray imaging. The umbilical cord consisted of a single artery and one vein. Multiple theories have been suggested for the pathogenesis of this condition, and despite recent progress in pathology, this condition remains debated.

Keywords: Mermaid syndrome; Sirenomelia; Single Lower Limb; Single umbilical artery.

INTRODUCTION

Sirenomalia or mermaid syndrome is an extremely rare and fatal congenital disorder with an incidence of 0.8 – 1 in 1,00,000 pregnancies . Male is to female ratio being 3:1.[1] The sirenomeliac reminds of the mermaid of Greek and Roman mythology, which was depicted as having the head and upper body of a human and the tail ofa fish.[2]

The most common feature seen in sirenomelia is the complete or partial fusion of the lower limbs into a single lower limb, giving it a mermaid resemblance. There are usually multiple underlying visceral abnormalities that make it incompatible with life with a very few rare exceptions of infants surviving with thiscondition.

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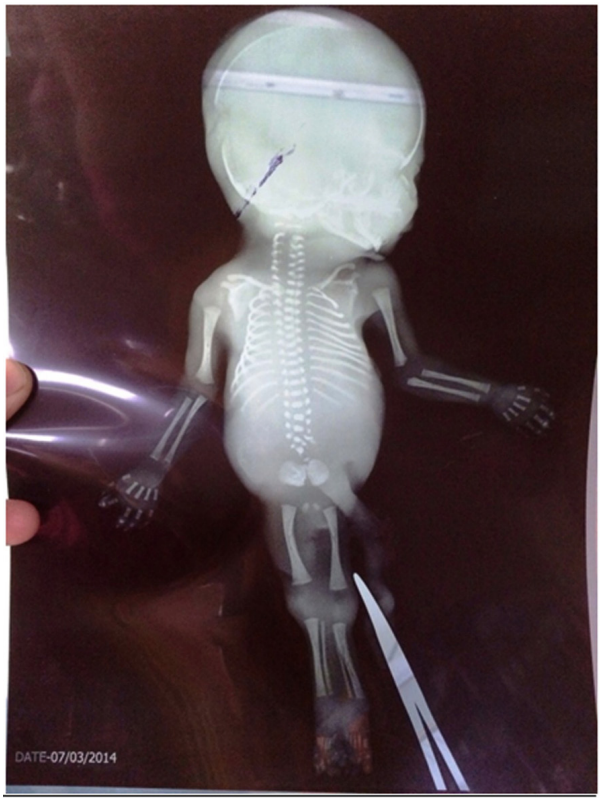
Here we have reported a case of aabortus with features of sirenomelia.

CASE REPORT

A 26 year old primigravida female came to hospital with severe lower abdominal pain and bleeding per vagina. She was immediately taken to the delivery room where she delivered an18 – 20 weeks old fetus of weight 480 gms and length 21cm.

On physical examination, the baby had fused legs. The umbilical cord examination revealed single artery and one vein. The external genitalia were absent and an imperforate anus was observed. An X – ray done revealed poorly expanded lungs and two distinct sets of femur and tibia.

Once the mother was stable and shifted to general ward, detailed history was taken, which revealed history of consanguineous marriage and history of tobacco use before pregnancy. But mother said she had not consumed tobacco once she knew she is pregnant. No any antenatal check up or previous ultrasonography was done. The patient was from a very low socio economic status and both husband and wife work as labourer. She had pain in lower abdomen on and off since 3-4 days and leaking PV since 1-2 hrs, so came to hospital.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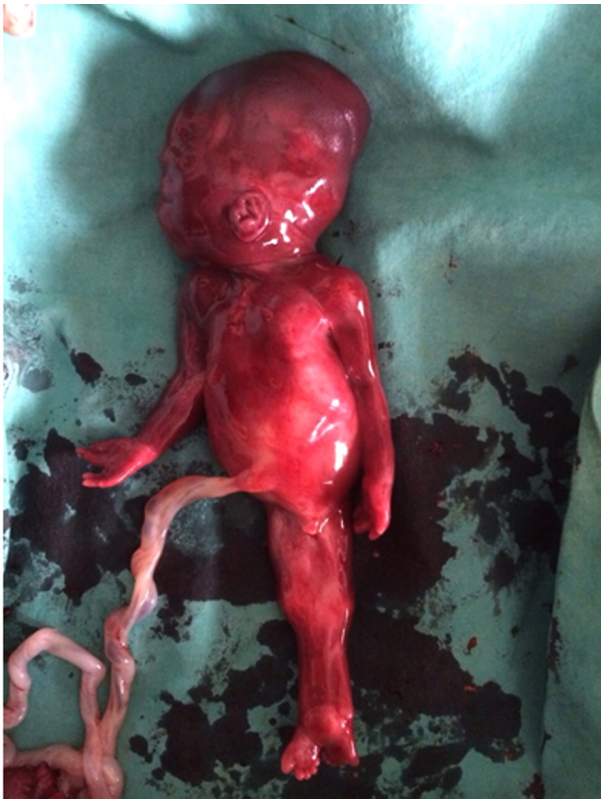

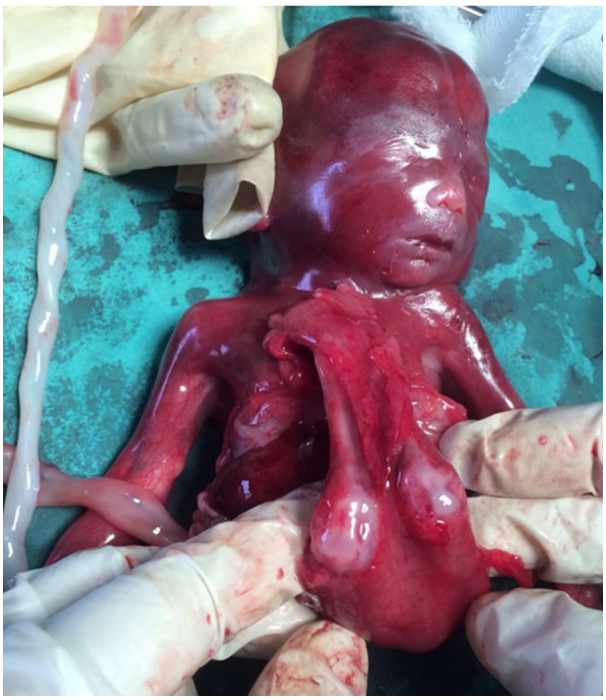

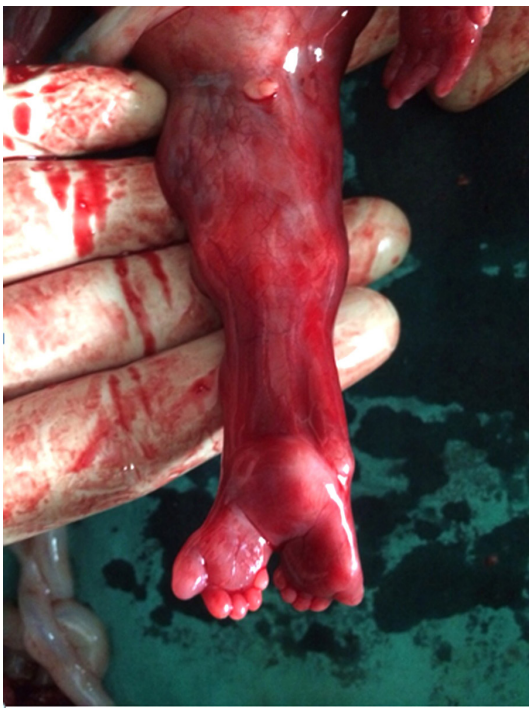

DISCUSSION

Sirenomelia (Mermaid syndrome) anomalies are the most severe form of caudal regression syndrome.[3] Caudal regression syndrome which is a rare congenital defect consists of a series of anomalies ranging from ectopic anus to sirenomelia, with a prevalence of 0.1- 0.2:10,000 in normal pregnancies in general population. But Prevalence is more (relative risk of 200 – 250) in diabetes.[4,5]

Various theories have been suggested for the pathogenesis of this condition but none is proved conclusive. This pattern of anomaly can be probably due to a generalised alteration of mesodermal cell migration that occurs between 28 to 32 days of gestation.[6] Altered oxidative metabolism may cause increase production of free oxygen radicals which may be teratogenic to the early stages of developing embryos. Mostly seen in uncontrolled diabetis in pregnancy.[7]

Another hypothesis of pathogenesis of mermaid syndrome in nondiabetic case has been proposed to be a vascular steal phenomenon with the single, aberrant, umbilical artery stealing blood supply from the lower torso and limbs. This leads to poorly perfused caudal region that udergoes partial or complete agenesis of caudal structures.[8,9] A possible vertebral dysgenesis leading to lowerlimb atrophyand inconsistent lower limb fusion can also be present.[10]

A case of sirenomelia without any arterial steal was published by Jaiyessimi et.al. indicating that other factors may also be involved in the pathogenesis.[11] The teratogenic agents like retinoic acid, cyclophosphamide, cadmium has been reported in the genesis of sirenomelia in animal studies.[12,13] Also there can be multifactor polygenetic transmission, dominant sex inked transmission and dominant autosomal transmission with a variable expression.[14]

CONCLUSION

Sirenomelia is a very rare and mostly fatal congenital anomaly. When diagnosed antenatally counselling and termination of pregnancy should be encouraged.

Regular antenatal checkup with optimum maternal blood glucose level, if diagnosed with gestational diabetis, should be maintained.

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