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Sirenomelia: A Fish Out of Water

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Abstract
Sirenomelia (Mermaid Syndrome) is a rare lethal multi-systemic birth malformation in which the two lower limbs are replaced with a rotated single midline tail-like limb. We are presenting a case of Sirenomelia(Mermaid Syndrome), which is an extreme example of Caudal Regression Syndrome. It invariably presents with lower limb fusion, sacral and pelvic bony anomalies, absent external genitalia, imperforate anus, and renal agenesis or dysgenesis. The baby passed away 1 hour after birth. It was small for gestational age. Young maternal age (21years) was the identified risk factor for sirenomelia in this case.

Keywords: Sirenomelia, Mermaid Syndrome, Caudal Regression Syndrome

1. Introduction:

Sirenomelia also known as mermaid syndrome is a fusion of lower extremities.[1] It is a rare and fatal congenital anomaly with an incidence rate of 0.8–4 per 60,000 to 100,000 pregnancies.[2,3] The exact etiologies are unknown, but maternal diabetes mellitus, teratogenic drugs, genetic susceptibility, vascular hypoperfusion, landfill water and young maternal age are known risk factors for this anomaly.[4,5] Even though sirenomelia manifestation may overlap with caudal regression syndrome (CRS) and VACTERL association, they are different entities.[6,7] Sirenomelia is more common in monozygotic twins and males.[8,9] We report a singleton baby with sirenomelia syndrome born to a teenage mother from West Bengal, India.

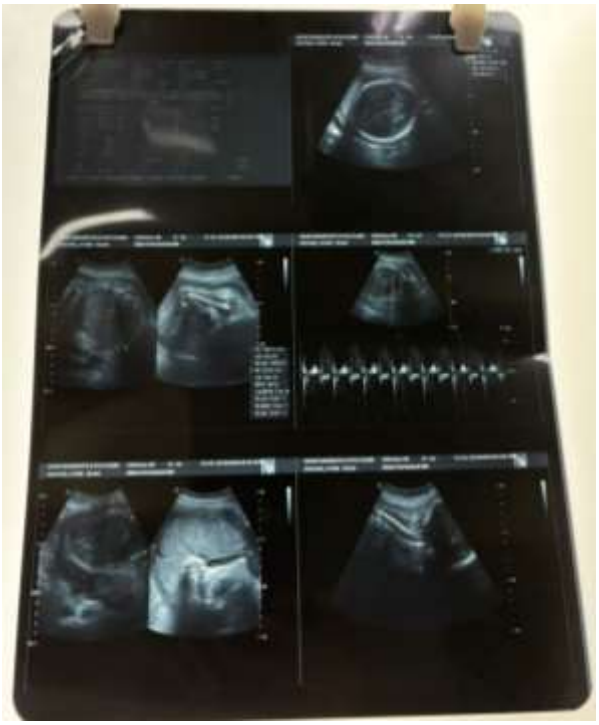
2.Case presentation:

History:

A 21 year old female presented to our hospital as a case of P $_{1+0}$ post vaginal delivery patient referred from nearby rural hospital due to severe oligohydramnios at 37 weeks of gestation. Per abdominal examination corresponds to 28 weeks of gestation with cephalic presentation. She was prepared for emergency LSCS due to severe oligohydraminos and fetal growth restriction and gave birth to a mermaid baby. The baby had fused lower limb with no visible external genitalia and anal opening. She is not a known case of GDM or PIH. Her 1st antenatal visit at 23 weeks of gestation and she started taking IFA thereafter. Anomaly scan reports as fetal parts could not be visualised properly due to very scanty liquor -advised for MTP but she continued her pregnancy. A total of 3 USG was done in antenatal period and patient had oligohydramnios

in all the scan. Both parents are in non-consanguinous marriage and in early 20's with no comorbidities. Their serology was negative(HIV1,2; HB_SAg; Anti HCV; VDRL) No history of alcohol intake or smoking or radiation exposure or genetic illness in the family.

Figure 1 : image of antenatal USG scan shows oligohydraminos



Physical Examination:

The new born was resuscitated for 10 minutes. The birth weight to gestational age was below the 10^{th} centile(Small for gestational age) on the Lubchencho curve. The baby had fused lower limbs and external genitalia were absent and cannot differentiate sex. There was no anal opening or dimple with a single umbilical artery. Upper part of the body was normal.

Figure 2: Shows fused lower limb

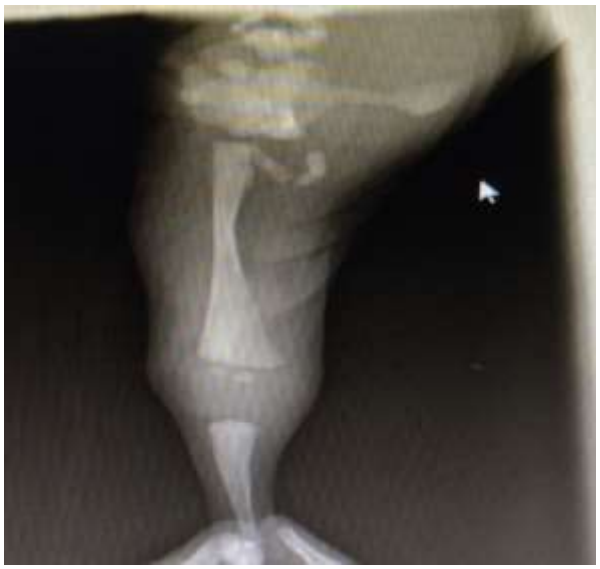
Figure 3 : Shows absence of external genitalia

Figure 4: shows absence of anal opening

Laboratory Test:

The newborn passed away 1hr after birth due to cardiac and respiratory arrest. Internal abnormalities could not be evaluated as the baby died soon. Newborn's x-ray of lower limb revealed single femur and single tibia (type VI sirenomelia according to Stocker and Heifetz classification).The mothers OGTT was normal. (HbA1c 5.5; FBS 85)The mother was discharged with no postpartum complications and with advice of birth spacing of minimum 1 year, mandatory antenatal visits, follow-up and periconceptional folic acid intake.

Figure 5: X ray shows single tibia and femur



Discussion:

Sirenomelia is an abnormal development of the caudal body of a fetus with partial or complete fusion of the lower extremity. It is also known as mermaid syndrome.[1,10] Sirenomelia is a multisystem severe malformation usually involving the gastrointestinal, genitourinary, cardiovascular, and musculoskeletal systems.[10] It is a rare and fatal congenital anomaly with an incidence rate of 0.8–4 per 60,000 to 100,000 pregnancies.[2,3] About 10%–15% of cases occur in twin births, most often monozygotic twins.[6] Sirenomelia is more common in monozygotic twins an. The risk of occurrence in one of two monozygotic twins is 100–150 times more than in singletons.[9,11]

The exact cause of sirenomelia is unknown. Researchers believe that both environmental and genetic factors may play a role in the development of this. Most likely, sirenomelia is multifactorial.[12]

Poorly controlled maternal diabetes is a known factor to be associated with sirenomelia. However, only ~0.5%–3.7% cases have been reported in diabetic mothers.[2] It is thought that increased free oxygen radicals in maternal diabetes, exert a teratogenic effect in embryonic development leading to this anomaly.[2,10,11] But in our case, there was no maternal diabetes mellitus.

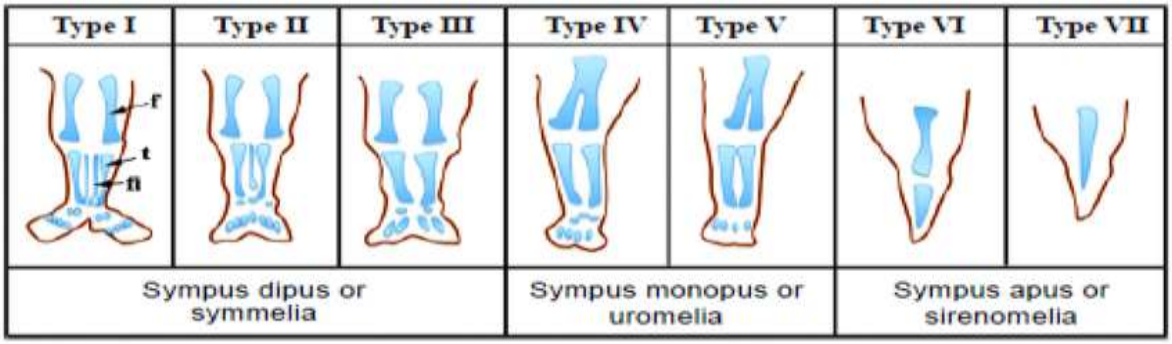
Although the primary molecular defect remains unknown, clinical studies have given rise to two main pathogenic hypotheses- these are the vascular steal hypothesis and the defective blastogenesis hypothesis.

The vascular steal hypothesis: According to the vascular steal hypothesis, fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn would result in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields. Vascular mechanisms have been proposed to underlie the pathogenesis of sirenomelia.[13]

The defective blastogenesis hypothesis: According to the defective blastogenesis hypothesis, sirenomelia is a primary defect of blastogenesis that occurs during the final stages of gastrulation at the tail bud stage, corresponding to the third gestational week in humans.[14]

Bmp signaling in caudal development and vasculogenesis: Bmp signaling performs multiple important roles during early embryogenesis, including the control of gastrulation and plays a crucial role in angiogenesis and vasculogenesis by promoting endothelial cell activation, migration and proliferation. Bmp signaling also directs maturation of the primitive capillary plexus into the mature vasculature.[15]

Stocker and Heifetz classified Sirenomelia infants from Type I to Type VII according to the presence or absence of bones within the lower limb.[16] Our case belongs to type VI category.



Conclusion:

Sirenomelia is a rare but fatal anomaly. When diagnosed antenatally, termination should be offered. Young maternal age is risk factors of this anomaly. A routine early second-trimester USG is recommended to make the diagnosis of sirenomelia. Patients who present with oligohydramnios along with fetal growth retardation either with or without history of leaking, we should look for fetal kidneys and bladder along with fetal vasculature. Therapeutic abortion can be carried out in earlier gestation due to invariably lethal condition. If diagnosed in later gestation we can avoid caesarean section for fetal indication. Lack of folic acid intake in the periconceptional period can lead to such congenital anomaly.

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