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SIRENOMELIA, THE FETAL MERMAID - A RARE CASE REPORT

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ABSTRACT Sirenomelia or mermaid syndrome is a rare congenital condition. Condition is characterised by fusion of lower limbs to form one single lower limb and is associated with bilateral renal agenesis, severe pulmonary hypoplasia, genitourinary abnormalities, anomalies of lumbosacral spine and pelvis, anorectal defect, gastrointestinal anomalies, single umbilical artery. It affects 1 in 100,000 live births worldwide, with the male to female gender ratio being 3:1. We report a case of sirenomelia type-III in a multigravida female reported to our hospital at term gestation. Her medical history was unremarkable, no associated risk factors, no history of drug abuse. No family history of congenital anomalies. Condition is fatal due to associated visceral anomalies. Prenatal diagnosis is difficult in late second and third trimester as severe oligohydramnios limits the visualisation of anomalies. Early diagnosis in first or early second trimester is recommended when the normal amount of amniotic fluid allow detailed sonographic scanning.

KEYWORDS : Oligohydraminos, Sirenomelia, Congenital anomaly, Renal agenesis, Potter's facies, Early Diagnosis

INTRODUCTION

Sirenomelia or mermaid syndrome is a rare congenital condition. It affects 1 in 100,000 live births worldwide, with the male to female gender ratio being 3:1[1]. Sirenomelia occurs with greater frequency in one twin of monozygotic twins than it does in dizygotic twins or singletons.[2] Its name originates from Greek word Siren meaning mermaid. Condition is characterised by fusion of lower limbs to form one single lower limb. It is associated with bilateral renal agenesis, severe pulmonary hypoplasia, genitourinary abnormalities, anomalies of lumbosacral spine and pelvis, anorectal defect, gastrointestinal defect, single umbilical artery.[3] It is often fatal during the new-born period. Etiopathogenesis of this disease is not clear, occur sporadically. Common risk factors include maternal drug (cocaine)abuse, retinoic acid, trimethoprim, maternal diabetes mellitus and consanguineous marriage.[3] There is no known association with chromosomal abnormalities. Here we present a case of sirenomelia type-III.

Case Report

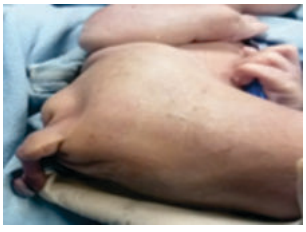
An unbooked Mrs. X 24 yrs. old G2P1L1 with 36 weeks + 4 days with previous LSCS with breech was admitted in labor room with complaint of pain abdomen. She had visited dispensary once for antenatal visit in 2nd trimester where she received tetanus booster. This was a spontaneous conception. She did not take any folic acid, calcium and iron tablets. She did not have any other significant trimester history. No past history of diabetes. No level -II scan was available. She had a single 3rd trimester scan at 30 weeks showing anhydraminos. She was married for 4 yrs. in a non-consanguineous marriage. In past, she had delivered a healthy male baby 2.5 yrs. ago at 40 weeks by lower segment cesarean section i/v/o fetal distress. Baby is alive and healthy. Patient did not have any antenatal and postoperative complications. There was no history of any drug (cocaine) abuse, retinoic acid, cadmium intake, no family history of any congenital anomalies.

Fetal sonogram was done on admission, it revealed single live intrauterine fetus with breech with anhydraminos with normal cardiac activity and dilated loops in abdomen, bladder was not visualized. All anomalies could not be ruled out due to anhydraminos. All blood investigations were normal.

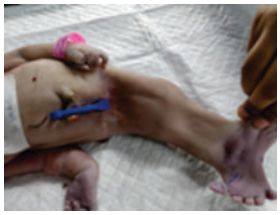
Emergency LSCS was performed i/v/o Previous LSCS with breech with anhydraminos in labor. She delivered a baby of 1.8kg who expired after 5 hours in NICU. On external examination of baby single umbilical artery was present. Baby had normal trunk and fused lower limbs up to ankles (Fig-3). Sex could not be identified as genitalia was absent. (Fig-3) No anal opening was seen. A tail like structure seen at caudal end approx. 4cm long (Fig-2). Right thumb was missing (reduction defect of upper limb). Deformity was present at right wrist (Fig-4). Both ears were deformed, low set ears, flattened nose, receding chin, suggestive of potter's facies. (Fig-1)

On USG- urinary bladder was empty and left kidney was not visualized suggestive of left renal agenesis. On x-ray-showed normal upper skeleton, single clavicle, deformed upper ribs, club foot, absent fibula, 2 femur, 2 tibia- sirenomelia type-III Due to religious reasons, parents did not give consent of autopsy.

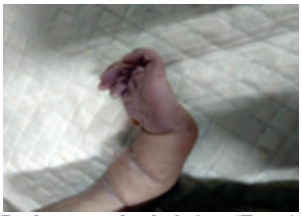
Potter facies (Fig-1)



Tail like structure (Fig-2)



Fused lower limbs and absent genitalia (Fig-3)



Right upper limb defect (Fig-4)

DISCUSSION

Sirenomelia or mermaid syndrome is a developmental defect that involve caudal region of body. It is diagnosed by fusion of the lower limbs and is associated with several internal visceral abnormalities. Various abnormalities present in sirenomelia include varying degree of fusion, anorectal atresia, vertebral

anomalies (sacral agenesis, ribs abnormalities), urinary tract anomalies (bilateral renal agenesis, ureteral, vesical and urethral agenesis), imperforate anus, absent urinary bladder, lumbosacral and pelvic bone abnormalities, single umbilical artery, and ambiguous genitalia.[2] Progressive oligohydramnios is usually the first sign of this syndrome in the second trimester because of renal abnormalities and is associated with pulmonary hypoplasia.[5]

This condition is fatal in about 60% cases because of bilateral renal agenesis and associated visceral anomalies.[5] Due to oligohydramnios, there is typical Potters syndrome which consists of potter's facies (low set ears, prominent epicanthic folds, hypertelorism, flat nose, receding chin, pulmonary hypoplasia). Prenatal diagnosis of sirenomelia has been made by ultrasound mostly during the first trimester, when the normal amount of amniotic fluid allows detailed sonographic scanning. Progressive oligohydramnios from 2nd trimester limits the visualisation of anomalies in later gestation.

Etiology of sirenomelia remain unclear but several theories have been proposed to explain the etiopathogenesis of sirenomelia-

a) Vascular Steal Hypothesis- Normally, the umbilical cord contains two arteries that originate from the iliac arteries, which return blood to the placenta. In cases of sirenomelia, the umbilical artery is single and arises from the abdominal aorta. The abdominal aorta distal to this branch directly bifurcates into iliac arteries without giving an origin to renal or inferior mesenteric artery branches. These vascular abnormalities lead to vitelline artery steal of the blood supply to the caudal end of embryo, these areas are underdeveloped and their growth is arrested in incomplete stage hence leads to sirenomelia and associated anomalies.[6]

b) Defective blastogenesis hypothesis- At blastogenesis, damage to the caudal mesoderm of the embryo between day 13 and day 22 of life results in merging, malrotation, and dysgenesis of the lower extremities.

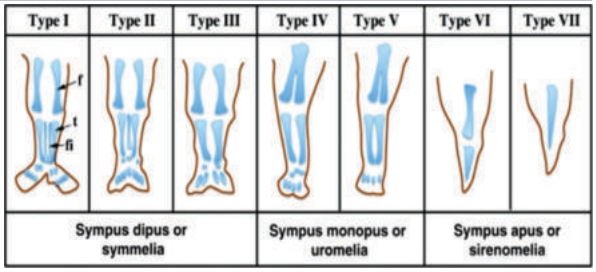
c) Oxidative metabolism from maternal diabetes may play a role as this can cause free oxygen radicals in the developing embryo leading to teratogenicity and mutation.[7]

d) Teratogens like retinoic acid, cadmium, and cyclophosphamide had been reported to cause sirenomelia in mice. However, no cases are reported in human beings.[8]

Currently there is no serum marker for sirenomelia to detect it antenatally. Early fetal scanning in first trimester is recommended to identify these gross anomalies, when the normal amount of amniotic fluid allow detailed sonographic scanning. Early diagnosis provides alterative management to the pregnancy.

In 2nd and third trimester due to oligohydramnios related to urinary tract agenesis or dysgenesis, it gets difficult to identify the malformations of the lower limbs.[9] In some cases, sirenomelia gets diagnosed only after delivery as seen in our case. Prognosis is very poor with babies being still born or passing away immediately after birth due to agenesis of the kidneys and pulmonary hypoplasia which are incompatible with life after birth.

Sirenomelia has been classified by Stocker and Heifetz 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.[10]



Classification of sirenomelia according to Stocker and Heifetz (Fig-5)

Other classifications, which focused on the degree of development of the fused legs denoted by the presence of feet has been abandoned. Sirenomelia used to be classified into three types according to the number of lower limb bones present –[11]

a) Sirenomelia Dipus: Two feet and two fused legs giving the appearance of a flipper.
b) Sirenomelia Unipus: One foot, two femurs two tibia, and two fibulas.
c) Sirenomelia Apus: No feet only one tibia and one femur.

Sirenomelia dipus, also called as Mermaid syndrome, has the most favourable outcome. Survival of children with sirenomelia depends on the associated visceral anomalies, especially renal function, rather than the sirenomelia itself.[12] In our report, foetus had sirenomelia type- III with left renal agenesis, single umbilical artery, absent genitalia, absent anal opening and potter facies. Diagnosis was made only after birth. In the ultrasound scan, the diagnosis was difficult because the first scan was done at later weeks of gestation and there was anhydraminos at that time.

CONCLUSION

Sirenomelia is usually always fatal due to renal agenesis and pulmonary hypoplasia. Early antenatal diagnosis gives parents the option to safely terminate the pregnancy early and to prevent the psychological effects associated with this condition.

Early scanning is recommended to identify these gross anomalies early in order to provide alternative management to the pregnancy. Ultrasound usually makes prenatal diagnosis during the late second trimester with the combination of malformation of the lower limbs and decreased amniotic fluid volume. But complete evaluation of a sirenomelia is hindered by associated severe oligohydramnios, making prenatal diagnosis difficult.

In contrast, amniotic fluid is rarely reduced in the first trimester thus making visualization of the fetus easier at this gestation. Hence, this condition can be diagnosed by early anomaly scan in late 1st or early 2nd trimester -presence of single immobile lower extremity, renal agenesis, absent bladder, single umbilical artery can be visualized. Fetal MRI has no constraints and can demonstrate various anomalies in greater detail than fetal sonogram, especially in late gestation.

All pregnant females should be registered and must have an early anomaly scan, folic acid supplementation, screening for diabetes, avoid teratogenic drugs, avoid cocaine/drug abuse in order to reduce the burden of congenital anomalies and its associated psychological harm to the mother.

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