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A Series of Sirenomelia (Apus, Unipus, Dipus)

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ABSTRACT

Introduction : Sirenomelia (Mermaid Syndrome) is a rare and lethal congenital anomaly. It has been considered as an extreme form of caudal regression syndrome Characterized by fusion of the lower limbs giving a characteristic mermaid-like appearance to the affected fetus. Commonly associated with Genito-urinary, Gastrointestinal, and Cardiovascular and Neural tube anomalies with an incidence of 1.5-4.2 per 100,000 births. There are approximately 300 cases reported in the literature, 15% of which are associated with twinning, most often monozygotic.

Series report : Here we are reporting three cases of different type of sirenomelia, symelia apus, symelia unipus, symelia dipus.

Conclusion : Prognosis of mermaid syndrome is very poor because of the condition involves variable major anomalies.

KEYWORDS : Sirenomelia, mermaid syndrome, Sirenomelia apus, Sirenomelia dipus, Sirenomelia unipus.

Introduction

Sirenomelia or the mermaid syndrome is a rare and lethal congenital malformation invariably associated with fusion of lower limb, sacral agenesis, anorectal malformation, genitor-urinary tract anomalies. The incidence of 1.5-4.2 per 100,000 births[1,2] in normal pregnancies which predominantly affect males. Oligohydromnios is universal and has strong association with maternal diabetes. The prognosis is very poor because this condition involves various major anomalies. Antenatal Diagnosis is possible with Ultrasound.

There are three variant of sirenomelia namely Symelia apus(No feet are present and the limbs are completely fused into a single limb), symelia unipus(partial fusion of both feet), symelia dipus(Two feet are present giving the appearance of fins).

Case one

Case one a 22-year-old primi gravida delivered a 1300gm stillborn baby (non-consanguineous marriage) at 32 weeks of gestation by

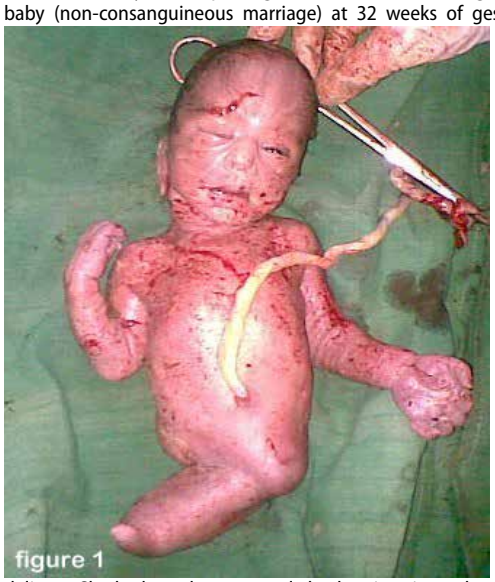
figure 1

delivery. She had regular antenatal checkup in primary health centre. There was no family history of congenital abnormality, No history of teratogenic drug intake or systemic illness like maternal diabetes mellitus. No Ultrasonography was performed. Patient refers to our hospital for premature rupture of Membranes. She eventually developed uterine contractions 6 hour later. Scanty Liquor was present at delivery. Examination of the infant revealed No feet are present and the limbs are completely fused into a single limb *(symelia Apus figure 1)*. Non pitting edema of hands, Facial edema, ear deformity, the pelvis was of small size, no anal opening and external genitalia were identified. The umbilical stump revealed no abnormality. Complete absence of genitourinary system was evident. The most important finding was pruned shape of the lower extremities.

Case two

A 26-year old primigravida (nonconsanguineous marriage) without any previous ultrasonographic screening of pregnancy status presented at term gestation. Caesarian section was done for fetal distress. This pregnancy was very poorly followed up few antenatal checkups. She had a normal antenatal course except poor progression of abdominal girth. On Clinical examination there was marked oligohydramnios which was associated with decrease fetal movements. Two year back she had a spontaneous abortion in the first trimester without any recorded complications. Her family history was unremarkable. A 2000gm, term baby of undetermined sex was born. The baby did not cry immediate after birth and required resuscitation to establish respiration. On examination Baby had both lower limbs fused into one and only one foot present which was inverted. Additionaly, the baby had flattened facies, absent genitalia (A small phallus like


figure 2

structure was seen in the back with an orifice), absent anal orifice and single umbilical artery .The fetus was diagnosed to be a case of *sire-*

nomelia unipus (figure 2). The face showed all the features of potter's facies with small pelvis. Cardiac clinical examination revealed ejection systolic murmur second left sterna border. Baby died within 2 hours.

Case three

A 23-year-old woman with non-consanguineous marriage delivered a baby 34 weeks of gestation by spontaneous vaginal delivery after an unsupervised pregnancy. She never had sonography screening. She had a normal antenatal course except poor progression of abdominal girth. The baby required bag and mask ventilation to achieve respiration. Baby had severe respiratory distress and died within 6 hours. No significant medical history and family history obtained except she was diabetic. On physical examination revealed a baby was 1900gm with Two feet are present giving the appearance of fins, the fusion of the limbs extends only as far as the ankle. The fetus was diagnosed to be a case of *symelia dipus (figure 3)*. The infant had potter facies, single umbilical artery, no anal opening and no discernable genitalia. Non pitting edema of hands was present. Complete absence of geni-

figure 3

Discussion

Sirenomelia, also known as Mermaid Syndrome, is a rare and lethal congenital malformation characterized by fusion of the lower limbs, in most cases, renal, vertebral, anorectal, or neural tube defects also exist with an incidence of 1.5 - 4.2 per 100,000 births[1,2] It is three times more common in males. Most of these newborns were still born or died immediately after birth; death is usually due to renal agenesis. Till date approximately 300 cases have been reported in the literature, Till 2006, 6 cases of surviving infants with mermaid syndrome were reported.

Exact cause of Sirenomelia is not known. Absence of chromosomal anomalies and familial inheritance has been noted in almost all cases. Oligohydramnios is frequent in mother. There is a strong association between this syndrome and maternal diabetes. However, only one of our cases had diabetic mothers. Incidence is higher in monozygotic twins [7]. In our case series twin gestation was not present in any of the cases. Various theories[3,4] have been postulated to explain the etiology of Sirenomelia. 1. Altered oxidative metabolism from maternal diabetes may cause production of free oxygen radicals in the developing embryo. 2. Vascular steal theory (blood is diverted from the caudal region of the embryo to the placenta producing a nutritional deprivation and abnormal development of caudal structures. 3. An embryological insult (Failure of caudal mesoderm blastogenesis). 4. External forces acting on the caudal extremity (mechanical compression by amniotic bands or oligohydramnios). 5. Unknown genetic mechanisms have also been postulated as causes of Sirenomelia. 6. As part of the caudal regression syndrome [8] (sacrum dysgenesis, spinal cord defect, urinary incontinence and misplaced lower limbs, renal dysgenesis and imperforate anus). 7. As part of VACTERL syndrome [6] (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities).

Sirenomelia is classified into three groups according to the number of feet present. The most common of the three conditions is **symelia apus**, in which both legs are merged completely into a single lower extremity, No feet only one tibia and one femur. **Symelia unipus** shows a presentation of one foot, two femora, tibiae and fibulae. In **symelia dipus**, two distinct feet are present but are malrotated and resemble fins. Our one case was compatible with symelia apus. Urogenital abnormalities are common including renal agenesis, absence of bladder and urethra, and absent or poorly formed internal and external genitalia, and imperforate anus. Only one umbilical artery may present. It is usually associated with other visceral defects such as hypoplastic lungs, cardiac agenesis, absent kidney, vertebral and central nervous system defects [5]. The fetuses with Sirenomelia have a Potter's facies (large, low-set ears, prominent epicanthal folds, hypertelorism, flat nose, and receding chin), as was found in our two cases. This is secondary to the renal abnormalities. Sirenomelia was formerly thought to be an extreme form of CRS; however it is reclassified to be considered a separate condition.

Antenatal diagnosis of sirenomelia is done by ultrasonography. However, in sirenomelic fetuses, bilateral renal agenesis causes severe oligohydramnios thus limiting ultrasound evaluation of the limbs in the second and third trimesters [9]. Though, in earlier gestational ages, the amniotic fluid volume may be sufficient to detect abnormal lower limbs. Additionally Colour Doppler imaging, 3D sonography and MRI can be helpful to find anomalies. Serum marker for antenatal diagnosis of sirenomelia is currently not available.

In our series however we could not establish the nature of the full foetal anomalies given that an autopsy was never performed. Besides, it was a poorly followed up pregnancy without any ultrasonographic evaluation. At delivery, clinical evaluation is usually sufficient to confirm the diagnosis. In our all cases, the diagnosis was obvious given the fused lower limbs anomalies, urogenital malformation, and imperforate anus. Sirenomelia carries with it a very poor prognosis. Survival is largely dependent on the extent of visceral anomalies, especially obstructive renal failure due to renal agenesis. Most of them die within few days. The management of sirenomelia is difficult and expensive, and the outcome is unpredictable.

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