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A Case Report Of Sirenomelia Sympus Dipus (Mermaid Syndrome)

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Abstract: *Sirenomeliasympusdipus or mermaid syndrome is a rare and fatal congenital condition, which is not associated with chromosomal anomalies. The fusion of the lower extremities characterizes it to form one single lower limb, sacral and pelvic bony anomalies, absent external genitalia, imperforate anus, and renal agenesis or dysgenesis and its etiology remains unclear. There are approximately 300 cases reported in the literature. This syndrome is incompatible with life due to the association of several congenital visceral abnormalities. Our case was a live born, normally delivered by assisted breech vaginal delivery at preterm by a 19-year-old second gravida of lower socioeconomic status. Examination of the baby revealed caudal dysgenesis having fusion of lower limbs. The extremities and both feet were fused with webbing of the toes and absence of external genitalia and anus. The baby died minutes after birth and we report this case due to its rarity and term live birth.*

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I. Introduction

Sirenomelia, or Mermaid syndrome, is a rare fatal congenital anomaly, characterized by replacement of normally paired lower limbs by a fused, single midline lower limb with a single umbilical artery, anal atresia and urogenital abnormalities.[2] Urogenital abnormalities result in oligohydramnios lead to characteristic Potter facies and pulmonary hypoplasia.[1] Features of Potter facies comprise flattened nose, low set ears, epicanthal folds, receding chin and micrognathia. Oligohydramnios is also the limiting factor in the prenatal diagnosis of sirenomelia by ultrasound. The incidence of sirenomelia is 0.98 per 1,000,000 births, and approximately 400 cases of sirenomelia have been reported to date.[2] Although sirenomelia is traditionally considered a uniformly fatal syndrome, there are a few rare cases where patients survived with the help of multiple surgeries for the variable period of the lifetime with a maximum up to the second decade.[2] Each of these rare cases documented the unilateral presence of one (or a partial) functioning kidney, and all had sympusdipus (fused lower limb with two feet) suggestive of less severe type. In the present case, we reported a case of sirenomeliasympusdipus in whichboth feet were fused with webbing of the toes.

II. Case Report

A 19-year-old adolescence with an unsupervised pregnancy at 30weeks' gestation was admitted with complaints of abdominal pain in Regional institute of Medical Sciences, Imphal, Manipur. On past history patient tells that she was gravida 2 and with history of abortion. She had no history of prior antenatal care and belonged to a tribal community of Manipur with lower socioeconomic status. There was history of exposure to chewing tobacco both before and during the pregnancy. There was no declared exposure to other drugs. She was otherwise healthy with no known history of genetic or congenital anomaly in her family. Ultrasonography of the case revealed a live foetus, in breech position, oligohydramnios and renal agenesis with severe caudal malformation, including fused lower extremities. The detailed evaluation was not possible because of the presence of severe oligohydramnios. The pictures of ultrasound were not included in this case report. At baby was delivered at 30 weeks of gestation with assisted breech vaginal delivery. The baby weighed 1500g with multiple congenital anomaly.The Apgar score was 3 at 1 min, 2 at 5 min and 0 at 10 min. The baby died within 20 min post birth in spite of resuscitation attempts by Paediatricians. On physical examination, the infant showed narrow chest, bilateral hypo plastic thumb with both feet were fused with webbing of the toes and absence of external genitalia and anus, umbilical cord with single umbilical artery (figure 1 and figure 2). The baby had typical Potters sequence facies including a flattened nose, prominent bilateral epicanthal folds, low set

ears and a receding chin (Figure 3). Autopsy was declined by the parents due to religious religion. Intrapartum and the postpartum period of mother was uneventful.

Figure:1

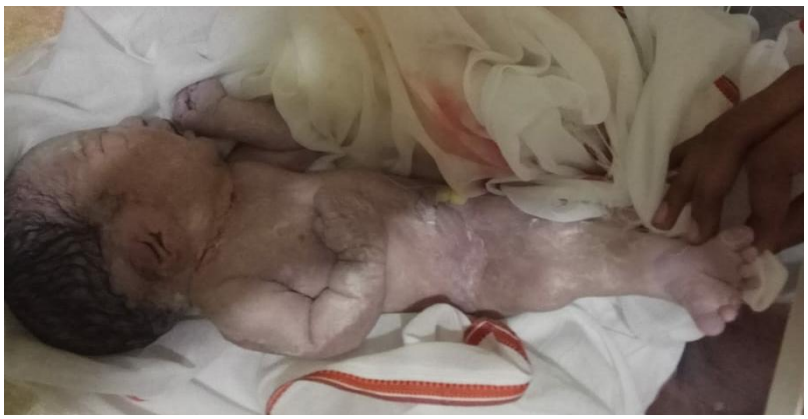

Figure:2

Figure:3

III. Discussion

Sirenomeliasympusdipus is a part of caudal regression syndrome but this has its own pathogenesis and exists as its own entity. Aetiology of sirenomelia remains unclear but it is known to be caused by an embryological injury that occurs between 28 and 32 days of development at the caudal mesoderm.[3]Sirenomelia is a rare congenital anomaly, till date approximately 300 cases have been reported in the literature of which 14 are from India. Most of these new-borns were still born or died immediately after birth; death is usually due to renal agenesis, which is incompatible with life. Swader reported the first surviving infant in 1989. Till 2006, 6 cases of surviving infants with mermaid syndrome were reported. The aetiology and pathogenesis of this malformation is unknown. Duhamel et al[4] in 1961 stated that sirenomelia and anorectal malformations represent the two extremes of a single comprehensive syndrome arising from an embryonal defect in the formation of the caudal region. He called it the syndrome of caudal regression

In a study by Stevenson et al[5] 11 cases of sirenomelia were examined and showed a pattern of vascular abnormalities. The common feature demonstrated was a single large abdominal artery which functions the umbilical arteries. The nutrients for this are diverted away from the caudal end of the embryo distal to the level of its origin. An early embryonic vascular network which supplies the yolk sac, has the steal vessel. It is formed from the vitelline artery complex and arteries below this vessel are underdeveloped, therefore any tissues formed from this vessel fail to have the nutrient supply thus leading to malformation of the lower extremities.[5]

Anomalies observed in sirenomelia are described as the most severe form of caudal regression syndrome. Fusion of lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia. Although the primary molecular defect resulting in sirenomelia is unclear, two main pathologic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis, fusion of the limbs results from a deficient blood flow and nutrient supply to the caudal mesoderm, which in turn results in agenesis of midline structures and subsequent abnormal approximation of both lower limb fields.[6] However in defective blastogenesis hypothesis, the primary defect in development of caudal mesoderm is attributed to a teratogenic event during the gastrulation stage. Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures. Maternal diabetes, tobacco use, retinoic acid and heavy metal exposure are possible environmental factors.[7]

Maternal diabetes is considered as a causative environmental factor for caudal dysgenesis because 10–15% of affected children have diabetic mothers.[8] In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or colour Doppler sonography.[9]The facial abnormality usually found in sirenomeliac infants known as Potter's facies, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. When features of Potter's facies are combined with oligamnios and pulmonary hypoplasia it is known as Potter's syndrome. Fifteen percent of patients with sirenomelia have associated twinning, which is most often monozygotic.[8]

Sirenomelia has been classified into three types according to the number of lower limb bones present:

a) Sirenomeliaapus: No feet only one tibia and one femur.
b) Sirenomeliaunipus: One foot, two femurs two tibia, and two fibulas.
c) Sirenomeliadipus: Two feet and two fused legs giving the appearance of a flipper.

Sirenomeliadipus has the most favourable outcome.[10]Survival of children with sirenomelia depends on the associated visceral anomalies, especially renal function, rather than the sirenomelia itself. Initial treatment of these new-borns includes supportive care and diverting colostomy, later management of these infants includes a multidisciplinary surgical approach involving various specialties.

Currently there is no serum marker for sirenomelia to detect it antenatally. Early scanning is recommended to identify these gross anomalies early in order to provide alternative management to the pregnancy. Ultrasound usually makes prenatal diagnosis mostly during the late second and third trimesters with the confirmation of combination of malformation of the lower limbs and decreased amniotic fluid volume, related to urinary tract agenesis or dysgenesis, these findings become visible after the end of the first trimester.[11] Like in our case, there are many cases with severe oligohydramnios reported in the literature, in which sirenomelia was diagnosed only following delivery of the baby. The first trimester prenatal diagnosis of the syndromes is very rare as because of the size of the foetus, which can make it challenging.[12]

IV. Conclusion

Sirenomeliasympusdipusis a rare fatal congenital malformation although less severe than sirenomeliaapus orunipus, severe visceral anomalies that decide the survival. 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. Early diagnosis of sirenomeliasympusdipus, gives the parents the option to safely terminate the pregnancy early in order to prevent the psychological effects associated with this condition. Patients who are at risk of this fatal condition should be offered early scanning to

correctly identify these anomalies to help counselling of the parents and prepare them for the outcome. Finally, this case illustrates the importance of accessing multidisciplinary team when a rare and unusual case presents.

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