Sirenomelia: The mermaid syndrome: A case report in Cambodia.

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Abstract

Sirenomelia (mermaid syndrome) is a rare congenital fetal anomaly with characteristic feature of complete or partial fusion of lower limbs. Although, this syndrome is incompatible with life due to the association of several congenital visceral abnormalities; however, there are few reports of surviving infants. Our first case was a shortly live born, normally delivered at 32 weeks by a 26-year-old primigravida mother of lower socioeconomic status. Examination of the baby revealed caudal dysgenesis having fusion of lower limbs, single leg with 1 foot and 3 toes. There was no identifiable external genitalia and anus. The infant died 5 minutes after birth and we report this case due to the rarity of abnormality, to report the prevalence of sirenomelia, and compare our findings with the literature.

Keywords: Caudal regression syndrome, mermaid syndrome, sirenomelia.

INTRODUCTION

Sirenomelia is a rare and fatal congenital defect characterized by fusion of the lower limbs and associated with some degree of lower extremity reduction. Patients also present severe malformations of the musculoskeletal system (e.g. sacral agenesis), as well as the urogenital and lower gastrointestinal tracts (e.g. renal agenesis, absent bladder, rectal/anal atresia, and absent internal genitalia). Most cases are stillborn, or die during, or shortly after, birth **[1].** The prevalence is about 1/1.000.000 births (http://www.orphanet.fr).

The incidence of sirenomelia is 0.8-1 case/100,000 births with male to female ratio being 3:1 **[2]**. The rarity of the case is obvious from the fact that many gynecologists might not have come across a case of sirenomelia in their whole professional carrier. There is a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of fetuses with this anomaly will have mothers with diabetes **[3, 4]**. We report a case of sirenomelia with no history of maternal drug abuse or maternal diabetes.

CASE REPORT

A preterm baby weighing 1.2kg was delivered vaginally at 32 weeks and 3 days of gestation by a 26-year-old primigravida with an unsupervised pregnancy. Postpartum investigation revealed the absence of diabetes mellitus. There was no history of drug intake and radiation exposure. The Apgar score was 5 at 1’ and 3 at 5 min following which the baby was died 5 minutes post birth due to severe respiratory distress. There was very rare amniotic fluid drained at the time of delivery. The new born baby had gross anomalies like narrow chest indicating lung hypoplasia, fused both lower limbs and one foot with 3 toes, absence of external genitalia, imperforate anus and single umbilical artery. Examination of the fused lower limbs showed the presence of all

thigh and leg bones thus classifying our patient as Type IV-V of Stocker and Heifetz classification (**figure 1**). The infant also had features of Potter's facies including prominent infraorbital folds, small slit-like mouth, receding chin, downward curved nose, and low-set ears, also the anomaly of left arm. Ultrasonography was difficult and impossible due to anamnios. Unfortunately, no autopsy and lower limb radiography were performed.

Figure 1: Classification Stocker and Heifetz.

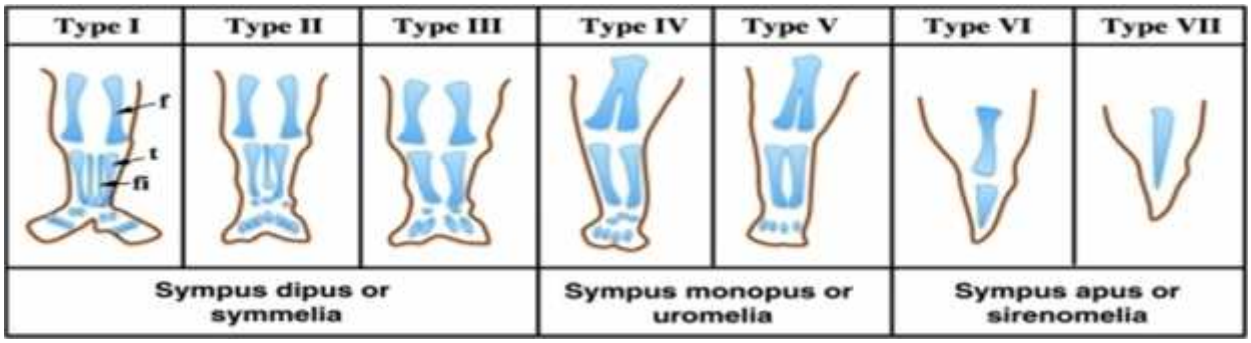

Figure 2: Sirenomelia baby with fused lower limbs containing 3 toes, Potter's facies, narrow chest, and absent external genitalia.

Figure 3: Anomaly of left arm

Figure 4: tiffany yorks.

DISCUSSION

Anomalies observed in sirenomelia are described as the most severe form of caudal regression syndrome **[5]**. Fusions of the lower extremities, presence of single umbilical and persistent vitelline artery are major features of sirenomelia **[6]**

Although the primary molecular defect resulting in sirenomelia remains unclear, two main pathogenic hypotheses namely the vascular steal hypothesis and the defective blastogenesis hypothesis are proposed. According to vascular steal hypothesis **[7]** 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. However in defective blastogenesis hypothesis **[8]** 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 **[9].** In our first case, there was no history of tobacco use before or during pregnancy. The maternal glycemia was normal.

Sirenomelia is usually fatal within a day or two of birth because of complications associated with abnormal kidney and urinary bladder development and function. In literature approximately 300 cases **[5]** are reported worldwide of which 14 are from India. In most of the cases the diagnosis was performed after birth. In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high resolution or color Doppler sonography **[10,11]**.The condition is usually incompatible with life due to visceral abnormalities especially that of renal system. Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks (**figure 4**), a girl who was born with fused legs. Over the years, she has undergone numerous operations to separate her lower extremities **[12].**

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. In our case, features of Potter's facies were present. When features of Potter's facies are combined with oligoamnios and pulmonary hypoplasia it is known as Potter's syndrome **[13]**. The left thumb was hypoplastic. Stocker and Heifetz classified Sirenomeliac infants from Type I to Type VII according to the presence or absence of bones within the lower limb **[15]**. Although we did not have radiographs to classify our case with certainty, nevertheless based on external examination, we suggest our first case belonged to Type IV-V (partially fused femurs and fused fibula).

CONCLUSION

Sirenomelia is a rare and lethal congenital anomaly. It is not only shocked for the parent but also for the practitioners who never cross this malformation in their professional lifetime. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal. Regular antenatal checkup with optimum maternal blood glucose level in pre conceptional period and in first trimester should be maintained to prevent this anomaly. Because this malformation is incompatible with life and the abnormality can be detected since first trimester, we recommend all sonographers take high attention to examine the extremities carefully and respect the recommendations giving by the ultrasound committee.

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