CASE REPORT

SIRENOMELIA: THE MERMAID SYNDROME

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ABSTRACT

Sirenomelia (the Mermaid Syndrome) is a rare and lethal congenital anomaly with an incidence of one in 100,000 of normal pregnancies. It is an extremely rare set of birth defects, which can vary in expression. The condition gets its name from one of the defects present, which is a fusing of the legs. The fused nature of the legs has been compared to a mermaid in appearance, hence the name. The characteristic features of Sirenomelia are complete fusion of the lower limbs, commonly associated with renal agenesis, absent external genitalia and other gastrointestinal defects. Another pathognomonic finding is the presence of single umbilical, persistent vitelline artery which is the chief distinguishing anatomic finding from Caudal Regression Syndrome. The termination of pregnancy is the choice of management as the case is diagnosed on the antenatal ultrasound which can show the features like renal agenesis with no liquor, fusion of the lower limbs. In our case report, the baby was delivered through Normal Vaginal Delivery with characteristic features of Sirenomelia on examination. The baby expired 8 hours after delivery.

KEY WORDS: Sirenomelia, Caudal regression syndrome.

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INTRODUCTION

Sirenomelia sequence or Mermaid Syndrome was originally described by Rocheus in 1542 and Palfyn in 1553 and named after the mythical Greek sirens.[1,2] It is a rare, lethal congenital anomaly characterized by fusion of the lower extremities or a single lower limb.[3,4] A number of other abnormalities such as genitourinary (absent external genitalia) and anorectal defects are frequently associated with this condition. The incidence of this syndrome is one in100,000 normal pregnancies.[1,6] This syndrome has a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22% of fetuses with this anomaly will have diabetic mothers.[6,7] We describe a case of Mermaid Syndrome diagnosed in the Neonatology Unit of Rehman Medical Institute, Peshawar, Pakistan after delivery.

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CASE HISTORY

A 34 years old G8P2 with previous 3 abortions and 3 child's death in the early neonatal period due to an unknown cause with history of cyanosis and sudden death, married since 12 years came to emergency department of Rehman Medical Institute, Peshawar in Labour pains since 4 hours. She was immediately referred to the Gynaecology ward for further and proper management. Her general physical examination revealed her to be B positive with hemoglobin of 9.8 gm/dl with blood sugar level of 86 gm/dl. There were no abnormality in the investigations; she was a 37 weeker with uterine contractions. Vaginal examination showed that the patient is in active labour with cervix dilatation of 6 cm. She had no significant past medical and surgical history. She remained in labour for 5 hours and then delivered a 1900 gram baby.

Gross examination of the baby displayed multiple external deformities. There was fusion of both lower limbs, absence of external genitalia with imperforate anus (fig 1 and 2). An emergency bed side ultrasound was performed that showed bilateral renal agenesis. The baby was given oxygen as he was not maintaining oxygen saturation and was kept in incubator and intravenous fluid was given but due to the underlying deformities the baby died 8 hours after birth.

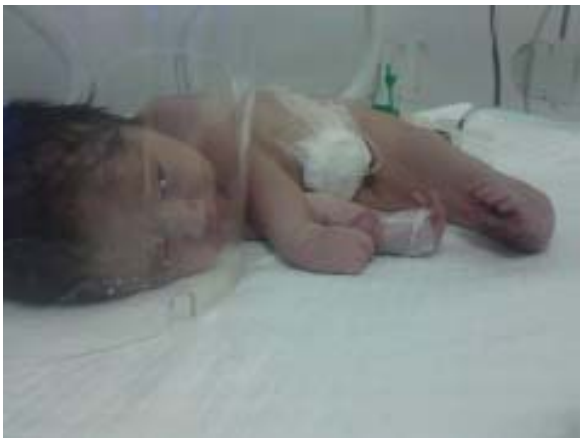

Figure 1: Photograph of patient with Sirenomelia

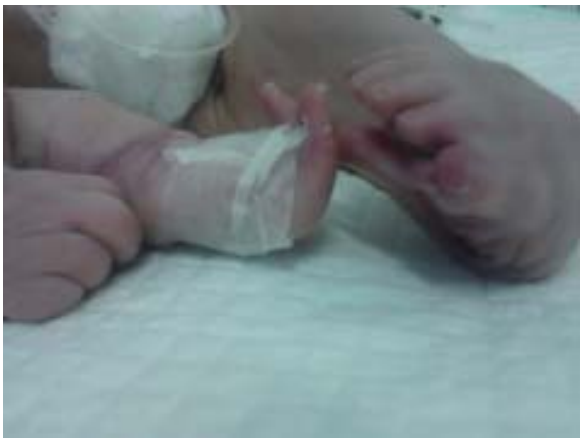

Figure 2: Close up photograph of patient with Sirenomelia

DISCUSSION

Sirenomelia is a rare congenital anomaly with an incidence of 1.5-4.2 per 100,000 births[8] with male predominance and higher incidence in identical twins, report indicates a 100-150 times higher incidence in monozygotic twins relative to dizygotic twins or singletons,[9] increase in risk with maternal age (less than 20 years and greater than 40 years).[8,10] Although it is a fatal conditions but there are reports of four children surviving with Sirenomelia.[10]

A number of risk factors are reported in different theories to be associated and one of the cause of mermaid syndrome is maternal diabetes.[11-13] Altered oxidative metabolism from maternal diabetes may cause production of free oxygen radicals in the developing embryo, which may be teratogenic,[14] however the case that we are reporting the mother was not diabetic. Genetic and environmental factors are also supposed to be the cause of mermaid syndrome. Teratogenic agents like cadmium elements, lead, vitamin A, cocaine and irradiation exposure are also reported to be associated with mermaid syndrome[15-17] The new reproductive technology such as ICSI (Intra Cytoplasmic sperm injection) has also some association with mermaid syndrome.[18]

Another hypothesis given by Stevenson et al explains that in mermaid syndrome, the blood is diverted from the caudal region of the embryo to the placenta producing a nutritional deprivation and abnormal development of caudal structures and the site at which steal occurs determine the severity of anomalies.[19]

Sirenomelia is a fatal condition because of bilateral renal agenesis which leads to oligohydramnios and lung hypoplasia. So far no treatment is available for Sirenomelia all we can do is to just prevent it either by avoidance of the exaggerating factors or by an early anomaly scan as it can be diagnosed as early as 13 weeks of pregnancy.[20,21] On second trimester ultrasonography the amount of amniotic fluid is sufficient to allow diagnosis and in third trimester diagnosis is impaired by severe oligohydramnios.

CONCLUSION

Sirenomelia is a rare and fatal congenital anomaly. Early prenatal diagnosis by first trimester scan should be the aim to diagnose it and minimize the trauma related to the termination of pregnancy at advanced gestation. In addition, where possible, a second US scan should be performed 4-6 weeks after the initial 8-9 weeks scan so that gross structural anomalies are detected and termination of pregnancy be considered earlier.

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CONFLICT OF INTEREST
Authors declare no conflict of interest.
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