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CASE REPORT

A Rare Case of Twin Gestation with Sirenomelia and a coexisting Normal Fetus

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ABSTRACT

Sirenomelia is a rare anomaly of caudal region of the body presented with fusion of lower limbs, in which genitourinary, gastrointestinal, cardiovascular, and neural tube anomalies are found in most cases. It is a very rare disorder with prevalence of 1 in 100,000 live births with a total of 300 cases reported till today in which 9 are from India. The precise etiology of sirenomelia was not well understood. Many theories have been proposed but none of these is considered definitive. Most of the times, the condition is fatal for the baby, even after surgery, and it is much more common in identical twins. This rare case in identical twins with one surviving fetus and other having fused foot with multiple congenital anomalies such as undetermined sex and renal agenesis is being reported.

Keywords: Fused foot, Renal agenesis, Sirenomelia, Stillbirth, Surviving fetus, Twin pregnancy, Two femurs, Two tibias.

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INTRODUCTION

Sirenomelia is a rare and fatal congenital anomaly characterized by single fused lower limbs with multiple urogenital and anorectal malformations with an incidence of 0.8 to 1 case/100,000 births, with male-to-female ratio being 3:1,[1] and cases have been reported from all ethnic groups worldwide.[2] Other anomalies associated with this condition are defects in lumbar and sacral vertebrae, renal agenesis, imperforate anus, and agenesis of internal genital structures except the testes and ovaries.

The term sirenomelia comes from "siren" or "mermaid" because of the characteristic fusion of the lower extremities. Because of its physical resemblance to the mythical mermaid, the topic of sirenomelia has fascinated the public for centuries. More than half the cases of sirenomelia result in stillbirth, and this condition is 100 times more likely to occur in identical twins than in single births or fraternal twins.[3] We present a case of identical twin gestation with one surviving fetus and the other stillborn having sirenomelia.

CASE REPORT

A 23-year-old primigravida with intrauterine pregnancy of 39 weeks came to our institution with pain in abdomen. She had sought regular antenatal care with transabdominal ultrasound done at 21 weeks and was diagnosed as a case of twin pregnancy with both live fetus of maturity 21 weeks 6 days. First fetus had normal parameters with normal spine, stomach, kidney, and four-chambered heart (Fig. 1). In second fetus, the lower limb had both thigh and lower leg fused with two separate femurs and two tibias and fused foot and renal agenesis, diagnosed as sirenomelia (Fig. 2). She had married life of 3 years, no family history of twins, diabetes, and malformations. On admission, she had mild pallor and no pedal edema, and pulse was 88/minute and blood pressure was 110/70 mm Hg. Per abdomen examination showed overdistended uterus with contractions, both fetal heart tones and first fetus in vertex presentation. Per vaginum examination showed os dilatation of 7 to 8 cm, full cervical effacement, and clear liquor, and she delivered in next half an hour. The placenta was diamniotic-monochorionic. First baby cried immediately after birth and weighed 2.5 kg. Second baby was 1 kg, did not cry 1 minute after birth, and had fused lower limbs and undetermined sex. Baby was shifted to neonatal intensive care unit but could not be revived. There was no postpartum hemorrhage. Consent of postmortem was not given due to ritual beliefs, so corpse was handed over to relatives. Postpartum period was uneventful, and mother was discharged on 3rd day with counseling. The first baby was found normal 1 month after follow-up.

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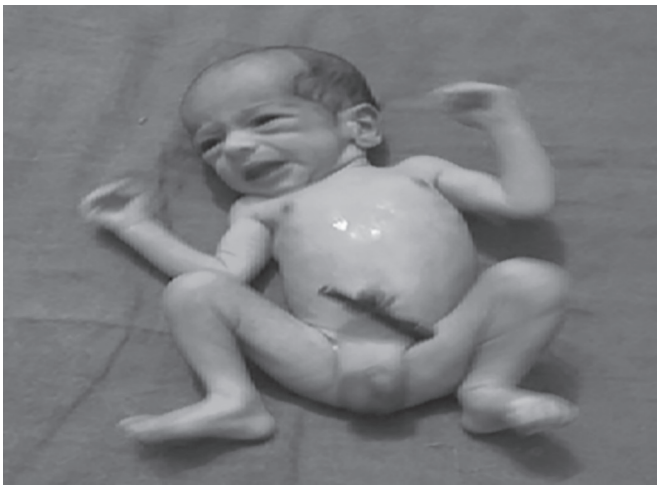

Fig. 1: Normal baby of the twins

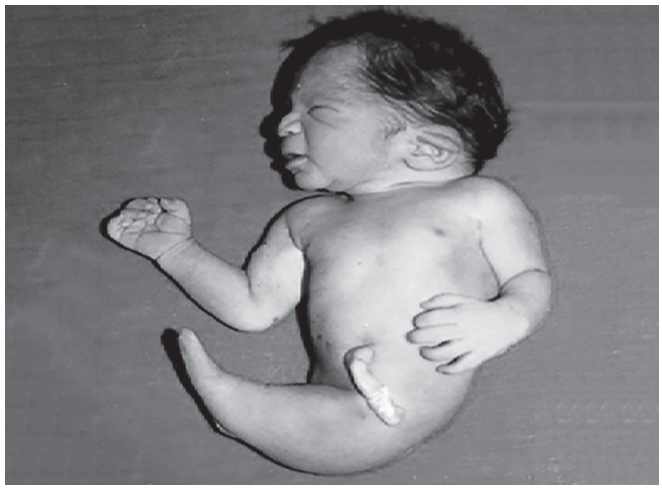

Fig. 2: Sirenomelic baby of the twins with fused foot

DISCUSSION

It is a rare and lethal congenital anomaly with unknown etiology showing abnormal development of the caudal region of the body involving varying degrees of fusion of the lower limbs with or without bony defects. Many different kinds of theories have been suggested such as maternal diabetes. However, our patient was not known to be diabetic.[4] Genetic and environmental factors are also noted. Hibelink et al[5] pointed out that teratogenic agents such as cadmium and lead may cause sirenomelia in the golden hamster. The likely potential teratogenic effect of vitamin A is mentioned by Von Lennep et al[6] with cocaine or irradiation exposure as well. Association of sirenomelia with new reproductive technologies, namely intracytoplasmic sperm injection, was also described.

Many theories have been proposed to explain its origin. Five pathogenetic theories of sirenomelia are described:

1. An embryological insult
2. Vascular steal theory
3. As part of the caudal regression syndrome (CRS)
4. As part of the VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, trachea-esophageal fistula, renal anomalies, and limb abnormalities)
5. External forces acting on the caudal extremity.

From an embryological point of view, the sirenomelia sequence comes from a caudal mesoderm damage taking place between days 28 and 32 of fetal life (first theory). Therefore, there are renal agenesis, lack of genital organs, imperforate anus, vertebral dysgenesis, and lower-limb atrophy.

Second theory states that sirenomelia is probably a consequence of an abnormal blood supply of caudal parts of the embryo. A diverged abnormal vessel carries blood through the umbilical cord into the placenta. The vessel "steals" blood from the structures located below its origin from the tissues of fetal caudal part. A third theory regards sirenomelia as part of the CRS, a rare congenital anomaly described by Duhamel, including sacrum dysgenesis, spinal cord defect, urinary incontinence, and misplaced lower limbs. Some inconstant features are renal dysgenesis and imperforate anus. Fifth theory is pressure theory stating that caudal hypoplasia is due to external forces, which is not well accepted. Cases of sirenomelia surviving beyond the perinatal period have been reported by Savader et al[7] for the first time in 1989 and later by Murphy et al[8] in 1992. However, in the majority of cases, the visceral anomalies are constant and uniform, incompatible with life as in our case. Survival is largely dependent on the extent of visceral anomalies, especially obstructive renal failure due to renal agenesis/dysgenesis as in our case.[9] The sirenomelia has been classified into Simpus Apus (no feet, one tibia, one femur), Simpus Unipus (one foot, two tibias, two fibula, two femurs), and Simpus Dipus (two feet, two fused legs) flipper like popularly known as mermaid. In our case, the baby had both thigh and lower leg fused with two separate femur and two tibial bones. The peculiar finding in our case was in twin pregnancy, which is very rare. More interestingly, the other twin was quite normal, thus excluding environmental factors, teratogenic drugs, and maternal diabetes from the etiological factors as both of the twins would had been equally exposed to predisposing factors. It could be a genetic aberration; however, karyotyping in these patients is always reported as normal.[10]

There have been reports of surviving sirenomelic fetuses. Pertinent among these is the case of the surviving infant with sirenomelia associated with absent bladder reported by Stanton et al.[11]

CONCLUSION

In our case, we could not find many of the features as described for this rare condition. Also, we could not further investigate to clinch the etiology due to noncooperation of the patient. This rare abnormality is usually universally fatal. This case was reported due to its rarity.

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