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CASE REPORT

Sirenomelia: A Rare Congenital Anomaly

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ABSTRACT

Introduction: Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body. The peculiar characteristics of sirenomelia are complete fusion of bilateral lower limbs giving the fetus an appearance of mermaid. The other anatomical defects may be renal agenesis, gastrointestinal defects, and absent external genitalia. The outcome of the condition is usually fatal for the baby despite attempts for corrective surgery. This abnormality was initially confused with caudal regression syndrome, but later was given a new name, i.e., sirenomelia mermaid syndrome.

Case report: We present a case of a 23-year-old primigravida unbooked case reported at 25 weeks 6 days period of gestation with complaint of decreased fetal movements. The ultrasound was suggestive of single live intrauterine fetus of 24 weeks with severe oligohydramnios (amniotic fluid index 1–2 cm). Based on fetal magnetic resonance imaging , which revealed multiple congenital anomalies including nonvisualization of kidneys and poorly formed lower limbs, provisional diagnosis of sirenomelia with renal aplasia incompatible with life was made. She delivered a baby 943 gm with features and appearance suggestive of mermaid syndrome (sirenomelia). Patient was discharged with advice to report early in next pregnancy.

Conclusion: Sirenomelia is a very rare disorder, with prevalence of 1 in 100,000 live births with a total of 300 cases reported until today in which 9 are from India. The precise etiology of sirenomelia is not well understood. Many theories have been proposed, but none of these is considered definitive. It is very important to diagnose this universally fatal condition by ultrasonography in early pregnancy, so that termination of pregnancy can be carried out.

Keywords: Caudal regression syndrome, Oligohydramnios, Sirenomelia.

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INTRODUCTION

Sirenomelia or mermaid syndrome is a rare and lethal congenital anomaly with incidence of 1 in 100,000 pregnancies. The rarity of the case is obvious from the fact that since the first case reported in 1542 AD, only 300 cases have been reported until now. Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body. The peculiar characteristics of sirenomelia are complete fusion of bilateral lower limbs giving the fetus an appearance of mermaid. The other anatomical defects may be renal agenesis, gastrointestinal defects, and absent external genitalia. An important finding that differentiates sirenomelia from caudal regression syndrome is the presence of single umbilical and persistent vitelline artery. The outcome of the condition is usually fatal for the baby despite attempts for corrective surgery. Sirenomelia is commonly associated with identical twins and has a still birth rate of almost 50%. We present a case of sirenomelia due to its rarity.

CASE REPORT

A 23-year-old female primigravida patient presented at 25 weeks 6 days gestation, referred to our center from peripheral hospital as a case of severe oligohydramnios. She was an unbooked case, and her first visit in gynecology outpatient department was at 25 weeks 5 days gestation with complaint of decrease fetal movements. Ultrasound report showed single live intrauterine fetus of 24 weeks, and severe oligohydramnios (amniotic fluid index of 1–2 cm). At the time of admission, her main complaint was inadequate perception of fetal movements. There was no history suggestive of rupture of membranes. There was no history of drug intake and radiation exposure. On examination, vitals were stable, mild pallor was present, per abdomen uterus was 24 weeks size with fetal parts easily palpable. She was admitted for evaluation and further management. Her hemoglobin was 8.5 gm%, viral markers and biochemical parameters were within normal limits. The fetal magnetic resonance imaging revealed multiple congenital anomalies, such as dolichocephalic skull, nonvisualization of kidneys, lordotic curve of lower spine, poorly formed lower limbs especially the legs not being clearly seen, and severe oligohydramnios. A provisional diagnosis of sirenomelia was made, and patient was counseled regarding the outcome of pregnancy. She

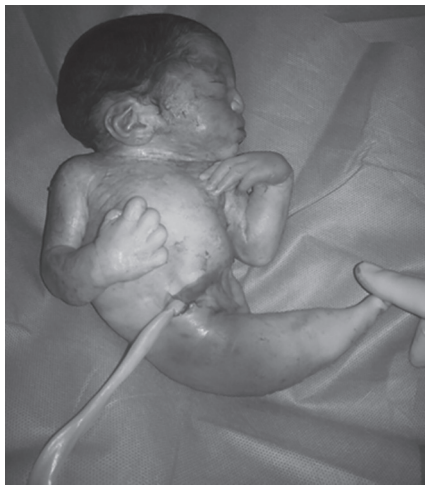

Fig. 1: Human siren reveals flexed single lower limb

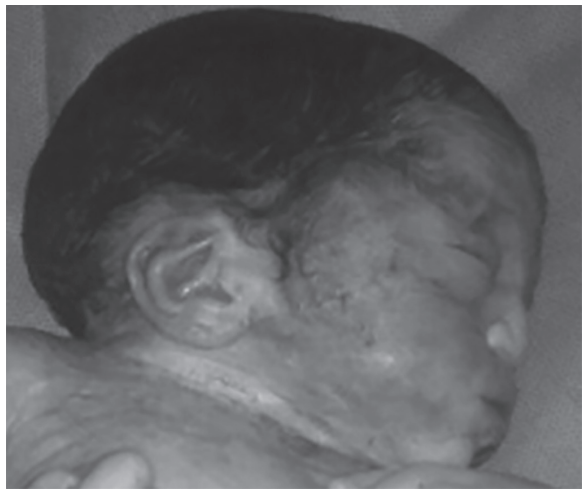

Fig. 2: Human siren with prominent infraorbital folds, small slit-like mouth, receding chin, downward curved nose, and low-set ears

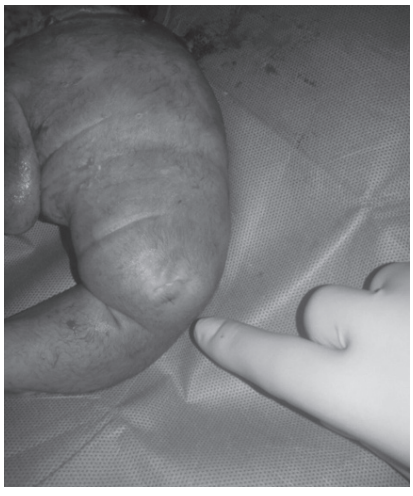

Fig. 3: Human siren with imperforate anus

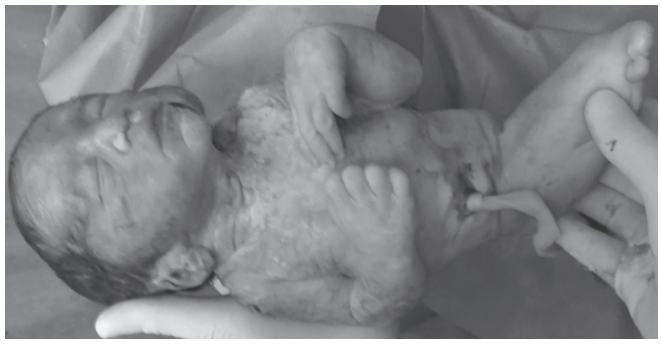

Fig. 4: Lack of external genitalia

had intrauterine fetal death and delivered a baby 943 gm with fused lower limbs (Fig. 1), prominent infraorbital folds (Fig. 2), small slit-like mouth, receding chin, downward curved nose, low-set ears, imperforate anus (Fig. 3), absence of external genitalia (Fig. 4), and single umbilical artery and appearance suggestive of mermaid syndrome (sirenomelia). The autopsy was refused by parents on religious grounds. Patient was discharged with advice to report early in next pregnancy.

DISCUSSION

A mermaid is a legendary aquatic creature with the upper body of a female human and lower half like fishtail. Mermaids are associated with the biological order *Sirenia* comprising dugongs and manatees. Hence, this syndrome is named sirenomelia. Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs.[1] It bears the resemblance of the mermaid of Greek mythology, and, hence, the synonym of mermaid syndrome.[2] The prevalence of this anomaly is 1:100,000 live births, with a male to female ratio of 3:1. About 300 cases have been reported in which 9 are from India.[3]

There are clinically mild and severe varieties. In the mild variety, the sirenomelia baby has two limbs fused into one, only to the extent of the skin. The feet may be fully formed and many are only attached at the ankles. All the three main bones of the leg are fully and correctly formed. In this situation, a small surgery can easily correct the deformity, whereas the severe variety is very difficult to manage. Externally, both limbs are completely joined and appear ill-formed. There is a complete absence of foot structures and out of the three long bones, only two are present in the entire limb. Other internal abnormalities can only be accessed with imaging studies. Sirenomelia has also been classified into three types: Simpus Apus (no feet, one tibia, one femur), Simpus Unipus (one foot, two femur, two tibia, two fibula), and Simpus Dipus [two feet and two fused legs (flipper like); this is called a mermaid].

The first case of sirenomelia was seen in 1542. Duhamel[4] gave the term syndrome of caudal regression in 1961, in which he described that sirenomelia and anorectal malformations represent two extremes of a single

comprehensive syndrome arising out of embryonal defect in the formation of caudal region associated with anorectal, genitourinary, and vertebral anomalies. Distinction was made between caudal regression syndrome and sirenomelia based on a specific pathogenic factor, namely arterial steal phenomenon. Stevenson et al[5] in 1986 dissected 11 cases of sirenomelia and postulated that these fetuses have a common feature of a steal vessel derived from vitelline artery complex. This single artery, connecting to aorta high in abdominal cavity, assumes the function of umbilical arteries and causes diversion of blood flow and nutrients from caudal portion of embryo to placenta, resulting in subsequent developmental arrest and malformations. The vessels distal to this aberrant umbilical artery were underdeveloped and malformed resulting in underdevelopment/malformed or developmental arrest in the incomplete stage of tissues dependent for nutrition on these vessels. An important finding that differentiates these two entities is presence of single umbilical and persistent vitelline artery. In antenatal period, sirenomelia can be diagnosed as early as 13 weeks by using high-resolution or color Doppler sonography.[6,7]

Ultrasonography of a fetus with sirenomelia demonstrates fused femur, decreased distance between two femurs, and decreased or absent mobility of the two lower limbs with respect to each other. When infant is clinically examined, there may be only simple fusion of skin of the limbs or there may be fusion of all long bones except fused femur. So, imaging studies, such as X-rays, which show the bony abnormalities, and ultrasonography are advised to see for solid organ abnormality. This disorder is universally lethal and, hence, prenatal diagnosis with imaging studies is very helpful to plan termination of pregnancy.[8,9] Exceptional cases without renal agenesis have survived, the best example being Tiffany Yorks, a 13-year-old girl who was born with fused legs. Over the years, she has undergone numerous operations to separate her lower extremities.

CONCLUSION

In our case, the external appearance and radiological findings were corroborative with this rare condition. Also, we could not further investigate to clinch the etiology due to noncooperation of the patient. This case was reported due to its rarity.

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