INTRODUCTION

Sirenomelia sequence is a very rare congenital anomaly with an incidence of one in 100,000 pregnancies. [1] It is also known as Sirenomelia, Mermaid Syndrome, Caudal regression, Caudal agenesis and Sacral agenesis. It is a birth defect of the lower body characterized by the apparent fusion of the legs into a single lower limb, giving them the appearance of a mermaid’s tail. This striking external phenotype is associated with a variable combination of severe visceral abnormalities, most commonly urogenital and gastrointestinal making Sirenomelia a multisystemic condition. The etiology of this is unknown. The condition may be the result of combined genetic and environmental component. The diagnosis is obvious at birth on examination of a baby, but pre-natal diagnosis can also be made as early as the first trimester by an ultrasound.

The etiology of this is unknown. Maternal drug use, Rheumatoid factor, maternal diabetes is identified as the common factors. In humans, Sirenomelia may be an autosomal dominant genetic condition and every single case is caused by a new spontaneous mutation. So the condition may be the result of combined genetic and environmental component. The important environmental risk factors described for caudal malformations are retinoic acid, maternal diabetes and heavy metals. Exposure to heavy metals is associated with Caudal dysgenesis and Sirenomelia in both experimental models and in humans. Maternal diabetes is a causative environmental factor for Caudal dysgenesis in 10–15% of affected children. However, this association remains controversial for Sirenomelia because only 0.5%–3.7% of reported cases has diabetic mothers [1,5].

CASE PRESENTATION

A 29-year-old, G4P3L3 unbooked, uninvestigated Hindu patient of lower socioeconomic status with unsupervised pregnancy of 34 weeks was admitted with labor pains in Bhagwan Mahavir Hospital, Pitampura, Delhi, India. History of the patient tells that she is gravida 4 and had previous history of three full term vaginal delivery. All three children were healthy and alive with no obvious congenital malformations. Until that date, the pregnancy was uneventful. Patient was a laborer in construction work and had a history of moderate exposure to tobacco. There was no declared exposure to other drugs. Patient had no antenatal ultrasonography report neither had any blood investigations. On clinical examination patient was of average built with mild pallor and pregnancy corresponding to 34 weeks of pregnancy in active labor. Fetal heart sound was audible with stethoscope. Consent for prematurity and risk of congenital anomaly taken and pt allowed to deliver vaginally.
Blood and urine investigations sent. Ultrasoundography could not be done as pt was in active labor. Patient delivered within one and half hour of admission an alive d malformed baby of 2100 gms vaginally. External genitalia were not distinct; hence sex of baby could not be declared on external examination. Intrapartum and immediate postpartum period was uneventful. Patient delivered within one and half hour of admission an alive d malformed baby of 2100 gms vaginally. External genitalia were not distinct; hence sex of baby could not be declared on external examination. Blood sugar level, hemoglobin A1C (HbA1C), urine for sugar of the mother were found within normal range and her hemoglobin level was 9.2 gm %. Examination of the baby revealed normal upper limbs but fusion of the entire lower limbs from the hip to just short of the ankle with bones present in the thighs (femur) and the legs (tibia and fibula) with bilateral talipes equinovarous deformity of feet. There was no anal opening and no discernable external genital organs (Figures 1 and 2). There was a spinal defect at the level of L2-L3. The umbilical stump revealed one umbilical artery and one vein. Baby shifted to neonatal intensive care unit (NICU) where the baby died after six hours. The parents refused autopsy of the baby so baby handed over to them. However, they allowed us to take an x-ray of the baby which showed radiological findings almost similar to this x-ray image available on web. (We could not upload our x-ray image because of poor image quality and non-availability of digital x-ray services.)

**DISCUSSION**

The cause of sirenomelia remains unclear as in our case. Our patient was non diabetic with previous normal pregnancies. Her tobacco addiction and working as laborer in construction works may have some environmental causal effect. There have been many theories about the etiology of the Mermaid syndrome. [5,6] Currently, there are two major theories described. According to Duhamel [7], Sirenomelia was the most severe form of congenital anomalies. He postulated that a small localized lesion would lead to anal imperforation and mild vertebral anomalies, larger lesions would lead to urinary tract and gastrointestinal malformations and lastly, extreme lesions would cause lower limb fusion and anomalies associated with Sirenomelia. Secondly, Stevenson [8] described an alternate theory of vascular steal that has recently taken favor over caudal regression for the Mermaid syndrome. Stevenson et al. explained that in the Mermaid syndrome, with the single vitelline umbilical artery blood is diverted from the caudal region of the embryo to the placenta producing a nutritional deprivation and abnormal development of the caudal structures. Most babies with sirenomelia have only one umbilical artery and one vein, as was seen in our patient.

Sirenomelia has been classified into three types according to the number of lower limb bones present: Sirenomelia apus: no feet only one tibia and one femur; Sirenomelia unipus: one foot, two femurs two tibia, and two fibula and Sirenomelia dipus: two feet and two fused legs giving the appearance of a flipper; also called as Mermaid syndrome. Sirenomelia dipus, which was the case in our patient, has the most favorable outcome. Survival of children with Sirenomelia depends on the associated visceral anomalies, especially renal function, rather than the Sirenomelia itself. Initial treatment of these newborns includes supportive care and diverting colostomy, later management of these infants includes a multidisciplinary surgical approach involving various specialties [9]. The diagnosis is obvious at birth on examination of a baby, but pre-natal diagnosis can also be made as early as the first trimester by an ultrasound [10,11]. This was not possible in our case as the mother’s pregnancy was unsupervised. After delivery, an infantogram can show the exact bony abnormalities while abdominal ultrasound can demonstrate abnormalities of the internal organs. An autopsy may clear the internal picture of babies with Sirenomelia. This was not performed in our case as the parents strongly refused any interventions.

**CONCLUSIONS**

To conclude Sirenomelia is a rare fatal congenital malformation of unknown etiology with severe visceral anomalies that decide the survival of fetus. Fusion of the lower limbs, which is very obvious, is less fatal. Few surviving patients need a multidisciplinary approach of treatment.
REFERENCES


