Case Report

A rare case of sirenomelia

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ABSTRACT

Sirenomelia also called mermaid syndrome is a congenital, rare, lethal, multisystemic human malformation of unknown etiology. The characteristic feature of sirenomelia is the fusion of the lower limbs, resulting in the appearance of a mermaid's tail, and thus the name "mermaid syndrome." This condition is also characterized by various severe urogenital abnormalities and the presence of a singular umbilical cord blood vessel, and it is more common in infants of diabetic mothers and in monozygotic twins. The incidence is around 1 in 60,000–70,000 pregnancies. The majority of affected fetuses are stillborn, whereas the rest of the live-born die in the early neonatal period due to complications of the gastrointestinal and urogenital systems. We are reporting a case of sirenomelia in a neonate born to an unregistered multipara mother. The baby had perinatal asphyxia, sirenomelia, dextrocardia, low set ears, lymphatic malformation, bilateral renal agenesis, absent external genitalia and anus, single umbilical artery, and congenital corneal clouding. There was no antenatal ultrasonography done and the baby died at 6 h of life.

Key words: Congenital malformation, Diabetic mother, Fusion of limbs, Mermaid syndrome

Sirenomelia, also popularly known as mermaid syndrome, is a rare lethal congenital deformity involving multiple systems with an incidence of approximately 1 in 60,000–70,000 pregnancies [1]. It was first described in the 16th century by Rocheus [2]. Duhamel in 1961 described all the anomalies of mermaid syndrome, including limb abnormalities, as the most severe form of caudal regression syndrome [2,3]. The characteristic feature of this syndrome is complete or incomplete fusion of lower limbs. It is also associated with renal abnormalities, oligohydramnios, absent external genitalia, single umbilical artery (SUA), imperforate anus, etc. [3,4]. Even though the precise etiology of sirenomelia is unclear, environmental trigger factor in combination with genetic predisposition is postulated [5,6]. As sirenomelia is a lethal perinatal condition, more than half of all affected cases are stillbirths [7]. <1% of babies survived beyond the 1st week after birth [4]. Those who survived longer than that expired within 1–2 years of birth due to urogenital system complications [8]. Recently, a few exceptional cases have survived owing to the presence of a functional kidney and reconstructive surgery to restore pelvic organs and separate the legs [9].

Access this article online	
Received - 07 April 2023 Initial Review - 19 April 2023 Accepted - 05 May 2023	Quick Response code
DOI: 10.32677/ijcr.v9i5.3993	

We are reporting a case of sirenomelia in a neonate born to an unregistered mother. The baby had perinatal asphyxia, sirenomelia, dextrocardia, low set ears, lymphatic malformation, bilateral renal agenesis, absent external genitalia and anus, SUA, and congenital corneal clouding. There was no antenatal ultrasonography done. The baby died at 6 h of life.

CASE REPORT

A 32-year-old unregistered G2P2L1A0 mother presented to the emergency room of our institute in labor at 36 weeks of gestation. The conception was spontaneous, and the marriage was non-consanguineous. There were no antenatal visits to the hospital, and no ultrasonography was done before presentation. There was no history of teratogenic drug intake, irradiation, or heavy metal exposure.

Her clinical evaluation on admission was suggestive of pregnancy-induced hypertension and gestational diabetes mellitus. The mother has a first female child who is normal and healthy. Our patient was delivered vaginally and had perinatal asphyxia and required intubation at birth. The baby was weighing 1560 g, length was 34 cm, and the head circumference was 32 cm. Anthropometry was suggestive of a small for gestation baby. The child had a single lower limb with no evidence of distal foot element. Other dysmorphic features

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included dolichocephaly, wide nasal bridge, bilateral corneal clouding, swelling in the neck, low-set ears with thin cartilage, and abnormal transverse crus in the ears, resulting in Stahl's ear appearance (Fig. 1). The child had a SUA, absent external genitalia, and anus. There was a redundant skin appendage similar to hypoplastic male genitalia present beside the single lower limb. The child had dextrocardia (Fig. 2).

Infantogram of the baby was suggestive of a single femur and an undeveloped tibia, with an absent fibula and there was no evidence of distal foot element. The neonate belonged to Type VI (sympus apus) according to the Stocker and Heifetz Classification [9]. Other prominent features noted on the X-ray were dextrocardia, absent pelvis, and L2 hemivertebra undeveloped tailbone (Fig. 3). Ultrasonography of the skull, neck, and abdomen showed an absent corpus callosum with a small posterior fossa, a multi-septated subcutaneous cystic lesion in the neck resulting from lymphatic malformation, non-visualization of both kidneys and gall bladder in the abdomen, and a splenic mass. The baby succumbed after 6 h of birth.

DISCUSSION

Sirenomelia is a lethal congenital anomaly characterized by the fusion of lower limbs, pelvic bone abnormalities, imperforate anus, absent external genitalia, and renal agenesis [10,11]. In a literature review, it is found that approximately 300 cases have been reported worldwide, among which 14 belonged to India [12,13]. The spectrum of malformation of the lower limbs seen in babies with sirenomelia ranges from the fusion of the skin of the lower limbs along the inner leg with fully formed and separate lower limb bones and fully formed feet which are fused at the ankles to the fusion of the legs into one lower limb with only two bones present in the entire limb (a femur and a tibia) and absence of foot structures [7]. This latter form was the case in our patient.

Although the etiology of sirenomelia is unclear, two pathogenic hypotheses are postulated. The first one is defective blastogenesis which involves the deficient mesodermal generation and subsequent impairment in the formation of caudal structures. According to this theory, sirenomelia could be considered a severe form of caudal regression [1,6]. Second hypothesis is an abnormal pattern of the vasculature. A normal fetus has two umbilical arteries with a normal origin; however, the majority of the cases of sirenomelia babies have a SUA with an abnormal origin derived from the vitelline artery [7]. Below the origin of SUA, the aorta becomes abnormally narrow, resulting in the shunting away of blood from the hypoplastic arteries supplying kidneys, gut, and genitalia, into the SUA. This causes the diversion of blood flow to the placenta, thus resulting in defective circulation and nutrition to lower limbs causing their arrested growth [1,6].

Maternal risk factors such as maternal diabetes, heavy metal exposure, teratogenic drugs (methylergonovine maleate, phenobarbital), irradiation, and caudal regression are well established in experimental studies and in humans [14]. In our case, the mother of the infant was diabetic which could have favored the disease. The occurrence of sirenomelia is 150 times the rate observed in dizygotic twin or singleton pregnancies [15]. Other risk factors such as like advanced maternal age and assisted reproductive technologies, namely,

Figure 2: (a) single-limb single with absent feet, neck lymphatic malformation, low set ears, and absent genitalia and absent anus; (b) dolicocephaly Stahl ear, rudimentary skin tag appearing like hypoplastic genatialia

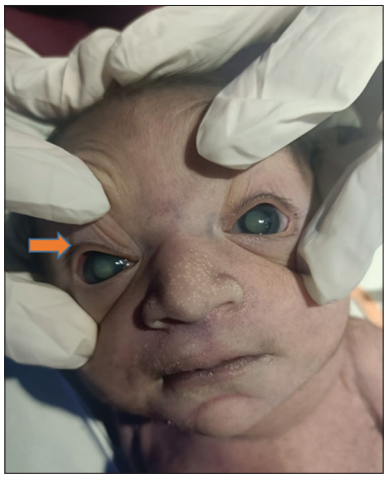

Figure 1: Bilateral congenital corneal clouding

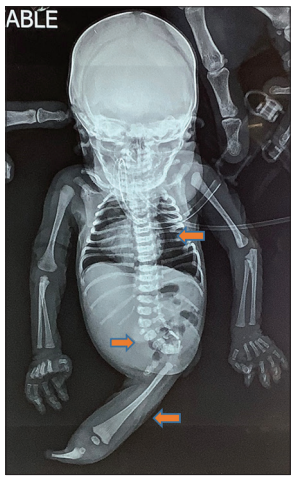

Figure 3: Infantogram showing dextrocardia, single lower limb with hypoplastic tibia and absent fibula, malformed pelvis

intracytoplasmic sperm injection have also been described [1,15] Unlike in humans, the genetic basis in the animal model is well established. A sirenomelia-like phenotype in animal models has been illustrated by genetic modifications through the induction of loss-of-function mutations in bone morphogenetic protein (Bmp) or gain-of-function mutations in RA signaling [1,6].

Other differential diagnoses that should be excluded are VACTERL/VATER (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities) connection, caudal regression syndrome, kidney malformations, and megacystis. Although sirenomelia is considered to be the most severe manifestation in the caudal regression syndrome spectrum, the two entities are separate nosologic disorders that require specific genetic counseling [16].

Sirenomelia can be diagnosed easily in first-trimester ultrasonography due to the presence of a sufficient amount of amniotic fluid that is less affected by fetal urinary production, than later in pregnancy. Fused lower limbs and oligohydramnios are key features that aid in the diagnosis during the first trimester of pregnancy [4,17]. Doppler ultrasound can be utilized to diagnose the presence of a SUA, especially in cases of oligohydramnios. A first-trimester ultrasound test should be done to minimize the physical and psychological trauma related to the termination of pregnancy at longer gestational periods [1]. In our case, the lack of antenatal visits for routine evaluation and presentation in the third trimester without any prior scans resulted in the diagnosis being missed, especially when the termination of pregnancy could have been feasible.

CONCLUSION

Antenatal ultrasound is of utmost importance in high-risk cases such as maternal diabetes, oligohydramnios, and exposure to heavy metals not only for the diagnosis of sirenomelia but also for other congenital malformations. The role of maternal diabetes has been highlighted in the incidence of sirenomelia. A combination of genetic and environmental factors could be responsible for the sporadic nature of this condition. In general, this condition is fatal in the perinatal period, and few surviving patients need a multidisciplinary approach to treatment.

ACKNOWLEDGMENT

We thank our Head of the Department Dr Surbhi Rathi for supporting us.

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Funding: Nil; Conflicts of interest: Nil.

How to cite this article: Gupta AM, Randad K, Singh VA, Virbhadre S, Ansari Q. A rare case of sirenomelia. Indian J Case Reports. 2023;9(5):125-127.