Case Report

Simpus Apus Sirenomelia – A case report of a rare congenital anomaly

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ABSTRACT

Sirenomelia, also known as "mermaid syndrome", is a rare congenital deformity of uncertain etiology. Sirenomelia is characterized by complete or partial fusion of lower limbs, giving the appearance of the tail of a 'mermaid'. This syndrome is almost always lethal due to associated congenital visceral abnormalities such as severe lung hypoplasia, and abnormalities of the kidneys, large intestines, and genitalia. We hereby, report a case of sirenomeila due to rarity of this condition and term live birth.

Keywords: *Caudal regression syndrome, Mermaid syndrome, Newborn, Potter's facies, Sirenomelia*

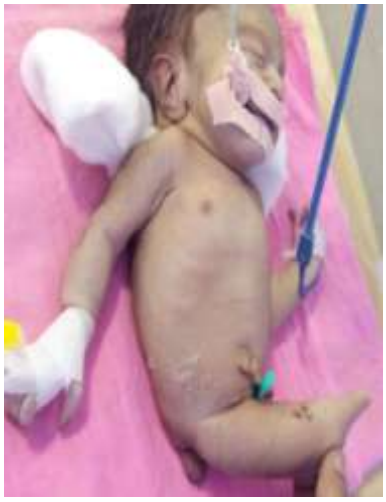
Sirenomelia is a rare and fatal congenital anomaly and more than half of the cases result in stillbirth. Sirenomelia is characterized by partial or complete fusion of lower limbs along with other associated malformations such as spinal anomalies, sacrococcygeal agenesis, genitourinary, and anorectal atresia [1]. Rocheus in 1542 and Palfyn in 1953 first described this sequence and named it after the mythical Greek sirens. In 1961, Duhamal described it as the most severe form of the caudal regression syndrome [2-3]. It results from a failure of normal vascular supply from the lower aorta *in utero*.

Reported incidence of sirenomelia is 0.8-1 case/100,000 live births with male bring predominantly affected (male to female ratio - 2.7:1) [4]. Its incidence increases 100-150 times in identical twins than in single births or fraternal twins. Maternal diabetes has been associated with caudal regression syndrome and sirenomelia and upto 22% of the fetuses with sirenomelia have mothers with diabetes [5-6]. We report a case of sirenomelia where maternal drug abuse and overt diabetes may have been the cause of this rare anomaly.

CASE REPORT

A preterm baby weighing 1.430 kg was referred to our hospital in view of respiratory distress and multiple congenital anomalies. Baby was delivered vaginally to a 28 year third gravid mother at 35 weeks of gestation. History of two first trimester spontaneous abortions was present, cause of which was not known to her and also no previous records were available. There was no history of prior antenatal care and she had not taken any medications including iron and folic acid tablets. Not antenatal ultrasonography was available with the parents. She belonged to a tribal community with lower socioeconomic status and history of tobacco chewing was present both before and during pregnancy. There was no history of diabetes mellitus, any illicit drug intake and radiation exposure. She was otherwise healthy with no known history of genetic or congenital anomaly in her family.

Baby was shifted to neonatal intensive care unit from another hospital and Apgar score was not known. At the time of admission, baby was hypothermic, with poor peripheral pulses and prolonged capillary refill time. Baby had severe respiratory distress; therefore, baby was put on



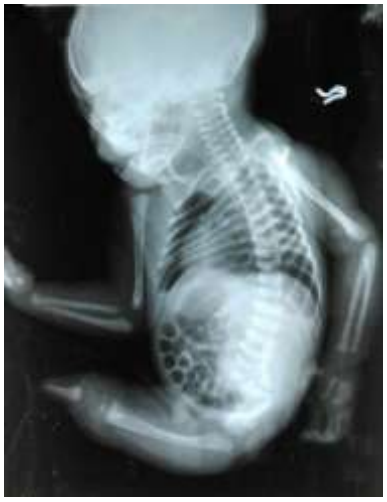


Figure 1: Clinical photograph of the baby showing single lower limb tapering downwards with no feet, absent external genitalia and a mass protruding from the anal region. Figure 2: Clinical photograph of the baby showing Potter's facies (prominent infraorbital folds, receding chin, downward curved nose, and low-set ears). Figure 3: Infantogram showing hypoplastic iliac bones, sacral agenesis, single femur bone and single rudimentary tibia.

mechanical ventilator. On physical examination, baby had gross anomalies like narrow chest indicating lung hypoplasia, small dysplastic pelvis and single lower limb tapering downwards with no feet. Also, absent external genitalia, imperforate anus and a soft tissue mass were seen protruding from the anal region, which had no identifiable structure and contained no bones (Fig. 1). The umbilical cord showed single umbilical artery. Facial deformities included prominent epicanthal folds, hypertelorism, downward curved nose, receding chin, and low-set ears and suggestive of Potter's facies (Fig. 2).

Radiological survey showed hypoplasia of the iliac bones, sacral agenesis, single femur bone and single rudimentary and hypoplastic tibia (Fig. 3). Therefore, this case was belonging to Stocker and Heifetz type VI. Ultrasound of abdomen to see the kidneys, and internal genitalia could not be done as ventilator parameters of the baby deteriorated and baby expired 10 hours after admission due to severe respiratory distress. Autopsy could not be performed as parents did not give consent for it.

DISCUSSION

Sirenomelia is a rare malformation affecting 0.1 to 1% of all malformed infants and is described as the most severe form of caudal regression syndrome. This deformity is also known as symmelia, symposia, sympus, uromelia, and monopodia. Caudal regression syndrome consists of a spectrum of anomalies ranging from ectopic anus to sirenomelia [2-3]. The sirenomeliac resembles the mermaid of Greek and Roman mythology, which was depicted as having the head and upper body of a human and the tail of a fish. This condition is incompatible with life and most of the sirenomelia cases result in stillbirths [4-6]. Though few survivors have been reported, such children rarely survive beyond the neonatal period [7-8]. The risk factors for sirenomelia include maternal diabetes, teratogens, genetic factors and maternal age less than 20 years [4-6].

The etiology of sirenomelia is still widely debatable and the primary molecular defect causing sirenomelia remains unclear. Two main proposed hypotheses are there namely the vascular steal hypothesis and the defective blastogenesis hypothesis. According to vascular steal hypothesis, abnormally formed umbilical cord blood vessels results in deficient blood flow and nutrient supply to the caudal mesoderm. In a normal foetus, umbilical arteries divide into two iliac arteries in pelvis and supply pelvic organs, genitalia and legs. However in sirenomelics, a single large artery assumes the function of umbilical arteries thus steals and diverts the blood flow and nutrition from caudal portion of the embryo to the placenta. This results in a small aorta and variable absence of the arteries affecting blood supply to the kidneys, large intestine, and genitalia, resulting in malformation of these organs [9-10].

On the other hand, defective blastogenesis hypothesis suggests that a teratogenic event such as maternal diabetes, tobacco use, retinoic acid and heavy metal exposure during gastrulation stage i.e. during 3rd gestational week may lead to primary defect in the development of caudal mesoderm [9-10]. Such defect interferes with the formation of notochord, resulting in abnormal development of caudal structures. Sirenomelia occurs in mice lacking Cyp26a1, an enzyme that degrades retinoic acid, and that develop with reduced bone morphogenetic protein signaling in caudal embryonic region. This discovery of the genetic basis in mice may be an important step toward the understanding of its pathogenesis. [9].

The typical malformation in sirenomelia consists of fusion of lower limbs which can be of varying severity, ranging from fusion of only overlying skin to the most severe form having only one lower limb tapering to a point with absence of foot structures. According to the presence or absence of bones within the lower limbs, Stocker and Heifetz classified sirenomelia into seven types (table 1) [11]. As evident from X-ray, our case belonged to Type VI (single femur, single tibia, and absent fibula).

Table 1 - Stocker and Heifetz classification of Sirenomelia

Type	Alternative terms	Characters
I	Simpus dipus or Symmelia	All thigh and leg bones are present
II		Single fibula
III		Absent fibula
IV	Simpus unipus or Uromelia	Partially fused femurs and fused fibulae
V		Partially fused femurs
VI	Simpus Apus or Sirenomelia	Single femur, single tibia
VII		Single femur, absent tibia

Sirenomeliac infants usually have Potter's facies, which includes large, low-set ears, prominent epicanthic fold, hypertelorism, flat nose and receding chin. Cardiovascular, respiratory and upper gastrointestinal tract malformations occur in 20-35% of cases. This anomaly has also been associated with lower spinal column defects, single or anomalous umbilical artery, upper limb, and central nervous system defects. Some cases may have radial agenesis, oesophageal atresia and tracheoesophageal fistula, which suggest VATER association. This is particularly seen in children of diabetic mothers [12]. However, no such correlation was found in our case. Association of sirenomelia with other very rare defects such as exstrophy of bladder, cyclopia/ holoprosencephaly, and acephalus acardia has also been reported [4,11].

Currently no serum marker is available to make antenatal diagnosis of sirenomelia. However, it can be diagnosed as early as 13 weeks of gestation by high resolution or color Doppler sonography. Convergent femoral bones that lie in a side - by - side configuration without change over time suggest the possibility of lower limb fusion. Suggested diagnostic clue on antenatal ultrasonography is the observation of fetal lower extremity fusion in the setting of oligohydramnios and bilateral renal agenesis [13-15]. An early antenatal diagnosis is very important in order to allow prenatal counselling for possible pregnancy termination as sirenomelia is incompatible with life. Regular antenatal check up with maintenance of optimum maternal blood glucose level in preconceptional period and in first trimester may help prevent this anomaly.

CONCLUSION

Although sirenomelia is usually associated with bilateral renal agenesis and absent urinary bladder, the present case has a mild hydronephrotic single kidney and partially filled urinary bladder. Sirenomelia is strongly associated with Potter`s syndrome and a diabetic mother, but this case was neither associated with a diabetic mother nor with Potter's syndrome. Here we highlighted some of the findings from associated literature. Sirenomelia is a rare and lethal congenital anomaly. When diagnosed antenatally, termination should be offered. However, prevention is possible and should be the goal.

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