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Case Report

Sirenomelia (mermaid syndrome): a rare lethal congenital anomaly – a case report from Northern India

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ABSTRACT

Sirenomelia/ Mermaid syndrome is an extremely rare, lethal congenital malformation, occurring at a frequency of between 1.5 and 4.2 cases per 100,000 pregnancies. It is characterized by varying degrees of fusion of the lower extremities and severe malformations involving the genitourinary and gastrointestinal systems. Its pathogenesis is not fully understood, with a multifactorial etiology involving genetic, teratogenic, and vascular factors. Depending on the associated malformations, death occurs in utero or within the first few days after birth. Prenatal diagnosis and imaging studies allow for reliable recognition and diagnosis. We report a case of sirenomelia diagnosed after birth, following a clinical examination of the newborn. A 24-year-old primigravida at 35 weeks of gestation presented in labor with transverse lie and markedly reduced amniotic fluid volume. Ultrasonography revealed anhydramnios along with absent end-diastolic flow on Doppler assessment. An emergency cesarean section was undertaken. A stillborn fetus weighing 1.8 kg was delivered, exhibiting fused lower limbs, single umbilical artery, ambiguous external genitalia, syndactyly of the right hand, and imperforate anus findings consistent with sirenomelia. Sirenomelia carries an extremely poor prognosis. Early identification through detailed antenatal imaging is essential for appropriate counseling and decision-making in affected pregnancies.

Keywords: Sirenomelia, Mermaid syndrome, Congenital malformation, Oligohydramnios

INTRODUCTION

Congenital malformations represent irreversible structural defects arising during embryogenesis. Sirenomelia, also termed mermaid syndrome, is among the rarest anomalies, with an incidence ranging between 1.5 and 4.2 per 100,000 pregnancies.[1] It is defined by partial or complete fusion of the lower extremities, frequently accompanied by visceral abnormalities affecting the renal, gastrointestinal and genitourinary systems.[2]

The condition occurs sporadically, with a male predominance and is considered distinct from caudal regression syndrome due to its unique vascular features.[3] The underlying pathogenesis is multifactorial, with hypotheses implicating abnormal vascular development and defective caudal mesoderm formation.[4] Affected fetuses commonly exhibit bilateral renal agenesis, absent urinary bladder, anorectal malformations and abnormal external genitalia. These anomalies contribute to severe oligohydramnios, which in turn results in pulmonary hypoplasia and poor perinatal survival.[5]

The precise etiology remains uncertain. However, two widely accepted hypotheses include the vascular steal phenomenon, where aberrant blood flow compromises

caudal embryonic development, and defective blastogenesis, involving disruption of caudal mesoderm formation during early embryogenesis.[6,7] We present a case of sirenomelia diagnosed postnatally, highlighting the challenges in antenatal detection and the importance of early anomaly screening.

CASE REPORT

A 24-year-old primigravida at 35 weeks of gestation was admitted to the labor unit with severe oligohydramnios. She had history of irregular antenatal visits. A previous ultrasound at 16 weeks had indicated severe oligohydramnios along with a single umbilical artery. At 19 weeks, the amniotic fluid index was recorded at 3 cm. Anomaly scan was not done. After this, she reported to our institute at 35 weeks gestation with transverse lie with severe oligohydramnios in USG.

On admission, repeat ultrasonography demonstrated- near absence of amniotic fluid (anhydramnios), inability to adequately visualize fetal anatomical structures , absent end-diastolic flow on doppler examination. The fetus was in transverse lie. In view of the obstetric indication and poor fetal condition, an emergency lower segment cesarean section was performed following administration of corticosteroids. A stillborn fetus weighing 1.8 kg was delivered. External examination revealed complete fusion of the lower limbs forming a single extremity, presence of a single umbilical artery, syndactyly involving the right hand, ambiguous genitalia, absence of a visible anal opening.



Figure 1: Stillborn fetus with single fused lower limb.

The placenta appeared morphologically normal on gross examination. There was no history suggestive of maternal diabetes, teratogenic exposure or consanguinity. Family history was non-contributory. The mother had an uncomplicated postoperative recovery.

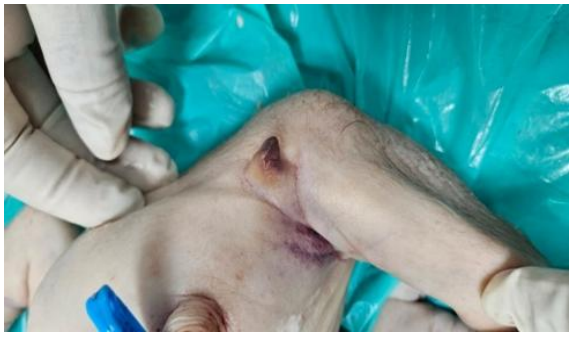


Figure 2: Sacral agenesis with single fused lower limb and imperforate hymen.

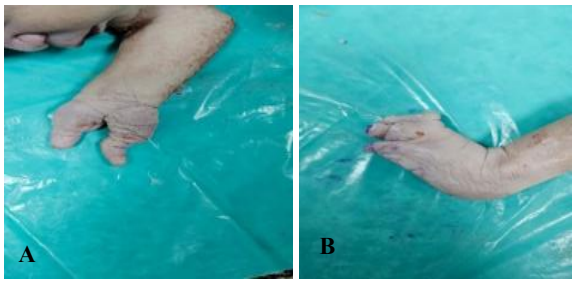


Figure 3: (A and B) Syndactyly in upper limb and lower limb.



Figure 4: X-ray of fetus showing sacral agenesis and single lower limb.

DISCUSSION

Sirenomelia is an exceptionally rare congenital anomaly involving the caudal region of the body and is frequently associated with life-incompatible visceral defects. Although sometimes considered within the spectrum of caudal regression, it is now regarded as a distinct clinical entity due to its unique vascular abnormalities.[6]

Pathogenesis

Two principal mechanisms have been advanced to explain sirenomelia are:

Aberrant vascular development (Vascular steal hypothesis): persistence of abnormal vitelline circulation may divert blood flow away from the caudal embryo, resulting in hypoperfusion and subsequent maldevelopment of lower body structures.[6]

Defective blastogenesis: disruption of caudal mesoderm formation during the third week of embryogenesis can lead to widespread anomalies involving the lower limbs, renal system, and gastrointestinal tract.[7]

Associated malformations

Commonly reported anomalies include: absent urinary bladder, imperforate anus, external genital abnormalities, vertebral and sacral defects and gastrointestinal malformations.[5,9] These abnormalities often culminate in severe oligohydramnios, which predisposes to pulmonary hypoplasia and markedly reduces neonatal survival.

Predisposing factors

Although most cases are sporadic, certain factors have been implicated such as Maternal diabetes mellitus, exposure to teratogenic agents, genetic susceptibility and environmental influences.[9] In present case, no identifiable risk factor was observed.

Prenatal diagnosis

Early diagnosis is feasible through targeted imaging modalities such as second trimester anomaly scan, color doppler evaluation and fetal MRI in selected situations. Typical antenatal findings include oligohydramnios, fused lower extremities, renal agenesisand a single umbilical artery.[10] However, severe oligohydramnios often limits visualization, particularly in later gestation.

Prognosis

The prognosis of sirenomelia is extremely poor. Most cases result in intrauterine demise or neonatal death due to pulmonary hypoplasia and renal failure. Rare survivors require complex, multidisciplinary care, but long-term survival remains exceptional.[3]

CONCLUSION

Sirenomelia is a rare and lethal congenital condition with profound structural abnormalities. This case underscores the critical role of early antenatal screening in identifying severe fetal anomalies. Prompt diagnosis facilitates informed counselling and appropriate management decisions.

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