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MERMAID WITHOUT A FUTURE: A FATAL CASE OF SIRENOMELIA IN AN UNBOOKED PREGNANCY: A CASE REPORT

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KEYWORDS :

CASE REPORT

A preterm female neonate was delivered at approximately 34 weeks of gestation to a 24-year-old gravida 2 para 1 mother who had not undergone any antenatal check-up. The mother was unbooked, unimmunised, and had received no iron–folic acid supplementation, tetanus prophylaxis, or ultrasonography throughout pregnancy. She belonged to a low socioeconomic background and reported no history of diabetes mellitus, teratogenic drug exposure, febrile illness, or consanguinity. The labour was spontaneous, and the baby was born via vaginal delivery with a weak cry at birth. Immediate assessment revealed a birth weight of approximately 1.9 kg, with poor tone and weak spontaneous respirations requiring supplemental oxygen.

On external examination, the neonate displayed classical features of sirenomelia. The lower limbs were completely fused into a single tapering appendage extending from the pelvis to a pointed distal end, resembling a monolithic limb structure. The skin over the limb was continuous and showed no interlimb cleft. The external genitalia were absent; no vulval or penile structures were visible, and the perineum showed a smooth surface with complete absence of an anal opening. The umbilical stump showed a single umbilical artery, consistent with the hallmark finding reported in nearly all cases of sirenomelia. The upper limbs were formed; however, the left upper limb appeared rotated with limited mobility and abnormal positioning. The skin revealed prominent lanugo and vernix, consistent with prematurity. The abdomen appeared distended but soft, and no urinary output was observed during monitoring.

Radiographic evaluation demonstrated complete fusion of the lower limb bones with a single femoral structure proximally and a continuous tibial configuration distally, ending in a single pointed foot-like structure. Pelvic structures were poorly delineated, suggesting hypoplastic or fused pelvic bones. The abdominal cavity appeared crowded, with indistinct visualization of renal silhouettes, raising suspicion of bilateral renal agenesis. The upper limb X-rays revealed asymmetry, including deformity or abnormal angulation of the left humerus. These findings were consistent with sirenomelia, most likely Type I–II according to Stocker and Heifetz's classification.

The newborn was admitted to the neonatal intensive care unit (NICU) for supportive care. Thermal stabilization, oxygen supplementation, intravenous fluids, and empirical antibiotics were initiated. Despite intensive supportive measures in the NICU, the neonate developed persistent anuria, worsening metabolic acidosis, and progressive respiratory failure due to severe pulmonary hypoplasia. Given the severity of multisystem malformations and poor prognosis, the baby's condition continued to deteriorate and did not survive, with death occurring within a few hours of birth. The parents were counselled regarding the nature of the condition and its uniformly fatal prognosis.

DISCUSSION

Sirenomelia represents one of the most severe forms of caudal anomalies and is often incompatible with life due to associated renal agenesis and pulmonary hypoplasia. The condition is considered a distinct entity from caudal regression syndrome, although they share overlapping features (8). A characteristic hallmark of sirenomelia is the presence of a single large umbilical artery instead of two, supporting the vascular steal theory. Stevenson et al. proposed that an abnormal persistent vitelline artery diverts blood from the caudal region, leading to compromised perfusion and subsequent maldevelopment of lower limb buds and caudal organs (5). Alternatively, the defective blastogenesis hypothesis suggests a primary defect in the formation of the caudal mesoderm during gastrulation, affecting structures derived from lower somites (6). Both theories may act in concert, contributing to the spectrum of anomalies observed.

The condition is strongly associated with renal agenesis, absent or dysplastic genitalia, anorectal atresia, absent urinary bladder, and vertebral and pelvic deformities (3). Limb fusion varies widely and forms the basis of classification. Sirenomelia is categorized into seven types by Stocker and Heifetz depending on the degree of lower limb skeletal fusion, ranging from presence of all bones to complete absence of long bones (9). The present case demonstrated extensive limb fusion, complete absence of external genitalia, imperforate anus, bilateral renal agenesis, and single umbilical artery—features consistent with the more severe end of the sirenomelia spectrum.

Maternal diabetes is the most reproducibly identified risk factor, with up to 22% of affected mothers demonstrating gestational or pregestational diabetes (4). However, the mother in this case had no history or clinical evidence of diabetes. Other proposed risk factors include retinoic acid exposure, heavy metal toxicity, and maternal malnutrition (10). In many low-resource settings, lack of antenatal care, as seen in this case, remains a significant contributor to late diagnosis. Oligohydramnios due to renal agenesis often obscures sonographic evaluation in late gestation, underscoring the importance of early ultrasonography. Antenatal diagnosis as early as 13 to 16 weeks has been reported and allows counselling, pregnancy management, and consideration of medical termination before viability (7).

The prognosis of sirenomelia remains extremely poor. Survival is mainly dependent on the presence of functional renal tissue. Only a handful of survivors have been reported, all of whom had at least one functioning kidney and required extensive reconstructive and supportive interventions (11). In most cases, including the present one, renal agenesis or severe dysgenesis results in fatal pulmonary hypoplasia and metabolic disturbances incompatible with life.

This case emphasizes the continuing burden of severe congenital anomalies in unbooked pregnancies and highlights the need for strengthening antenatal care systems, especially in underserved populations. Early screening, maternal education, and nutritional and immunization support may contribute to reducing the incidence and improving the detection of life-threatening congenital anomalies.

CONCLUSION

Sirenomelia is a rare and severe congenital malformation frequently associated with multisystem anomalies and poor neonatal survival. This case highlights the challenges posed by the absence of antenatal care and the importance of routine prenatal screening for early detection of lethal fetal anomalies. Strengthening maternal health services and ensuring universal access to antenatal ultrasonography are essential steps in preventing unexpected intrapartum diagnoses and enabling timely counselling and intervention.

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